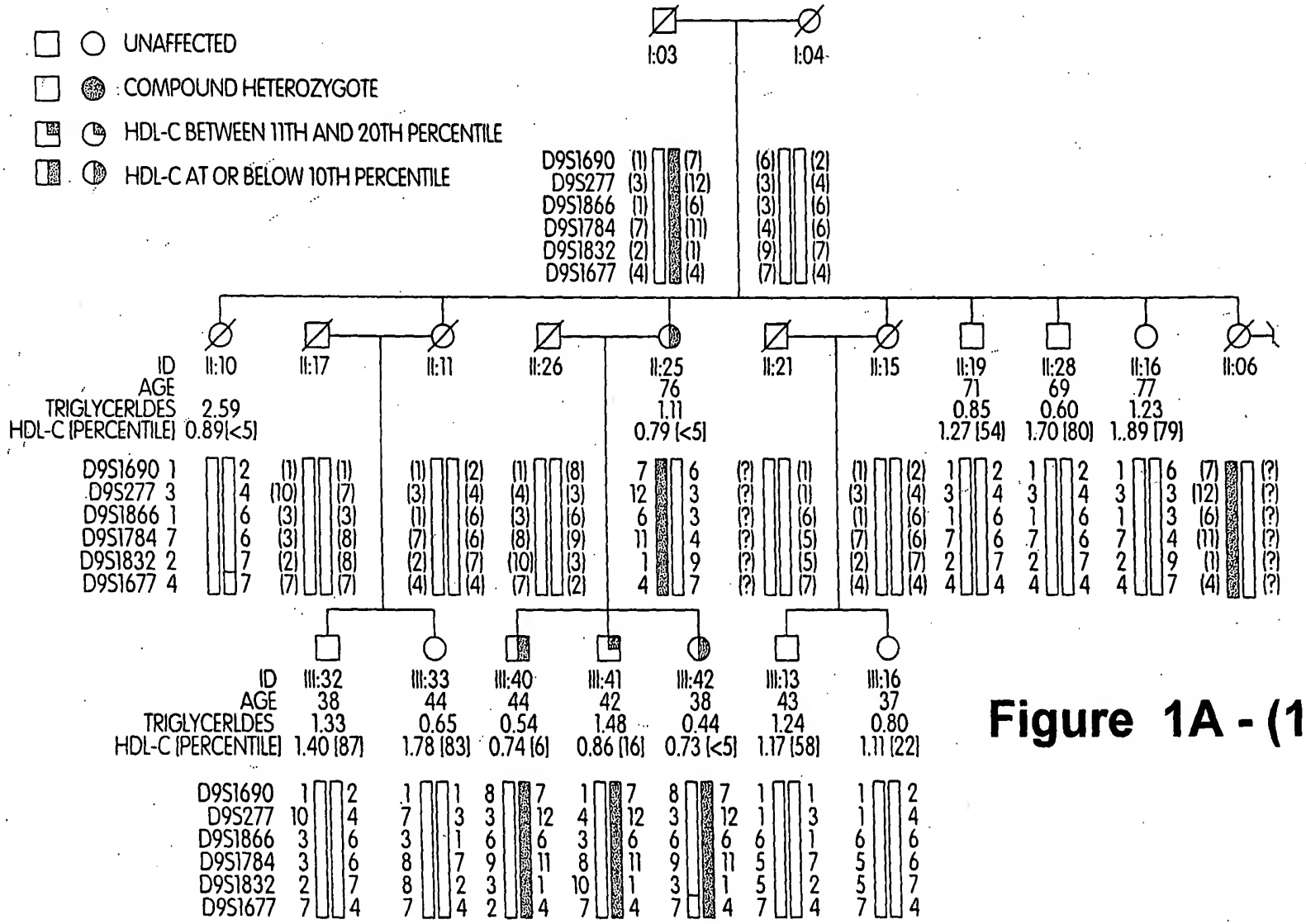
UNAFFECTED
COMPOUND HETEROZYGOTE
HDL-C BETWEEN 11TH AND 20TH PERCENTILE
HDL-C AT OR BELOW 10TH PERCENTILE
I:03
I:04
D9S1690 (1) (7) (6) (2)
D9S277 (3) (12) (3) (4)
D9S1866 (1) (6) (3) (6)
D9S1784 (7) (11) (4) (6)
D9S1832 (2) (1) (9) (7)
D9S1677 (4) (4) (7) (4)
ID II:10 II:17 II:11 II:26 II:25 II:21 II:15 II:19 II:28 II:16 II:06
AGE 76 71 69 77
TRIGLYCERLDES 2.59 1.11 0.85 0.60 1.23
HDL-C (PERCENTILE) 0.89(<5) 0.79 (<5) 1.27 (54) 1.70 (80) 1..89 (79)
D9S1690 1 2 (1) (1) (1) (2) (1) (8) 7 6 (?) (1) (1) (2) 1 2 1 2 1 6 (7) (?)
D9S277 3 4 (10) (7) (3) (4) (4) (3) 12 3 (?) (1) (3) (4) 3 4 3 4 3 3 (12) (?)
D9S1866 1 6 (3) (3) (1) (6) (3) (6) 6 3 (?) (6) (1) (6) 1 6 1 6 1 3 (6) (?)
D9S1784 7 6 (3) (8) (7) (6) (8) (9) 11 4 (?) (5) (7) (6) 7 6 7 6 7 4 (11) (?)
D9S1832 2 7 (2) (8) (2) (7) (10) (3) 1 9 (?) (5) (2) (7) 2 7 2 7 2 9 (1) (?)
D9S1677 4 7 (7) (7) (4) (4) (7) (2) 4 7 (?) (7) (4) (4) 4 4 4 4 4 7 (4) (?)
ID III:32 III:33 III:40 III:41 III:42 III:13 III:16
AGE 38 44 44 42 38 43 37
TRIGLYCERLDES 1.33 0.65 0.54 1.48 0.44 1.24 0.80
HDL-C (PERCENTILE) 1.40 (87) 1.78 (83) 0.74 (6) 0.86 (16) 0.73 (<5) 1.17 (58) 1.11 (22)
D9S1690 1 2 1 1 8 7 1 7 8 7 1 1 1 2
D9S277 10 4 7 3 3 12 4 12 3 12 1 3 1 4
D9S1866 3 6 3 1 6 6 3 6 6 6 6 1 6 6
D9S1784 3 6 8 7 9 11 8 11 9 11 5 7 5 6
D9S1832 2 7 8 2 3 1 10 1 3 1 5 2 5 7
D9S1677 7 4 7 4 2 4 7 4 7 4 7 4 7 4

Figure 1A - (1)

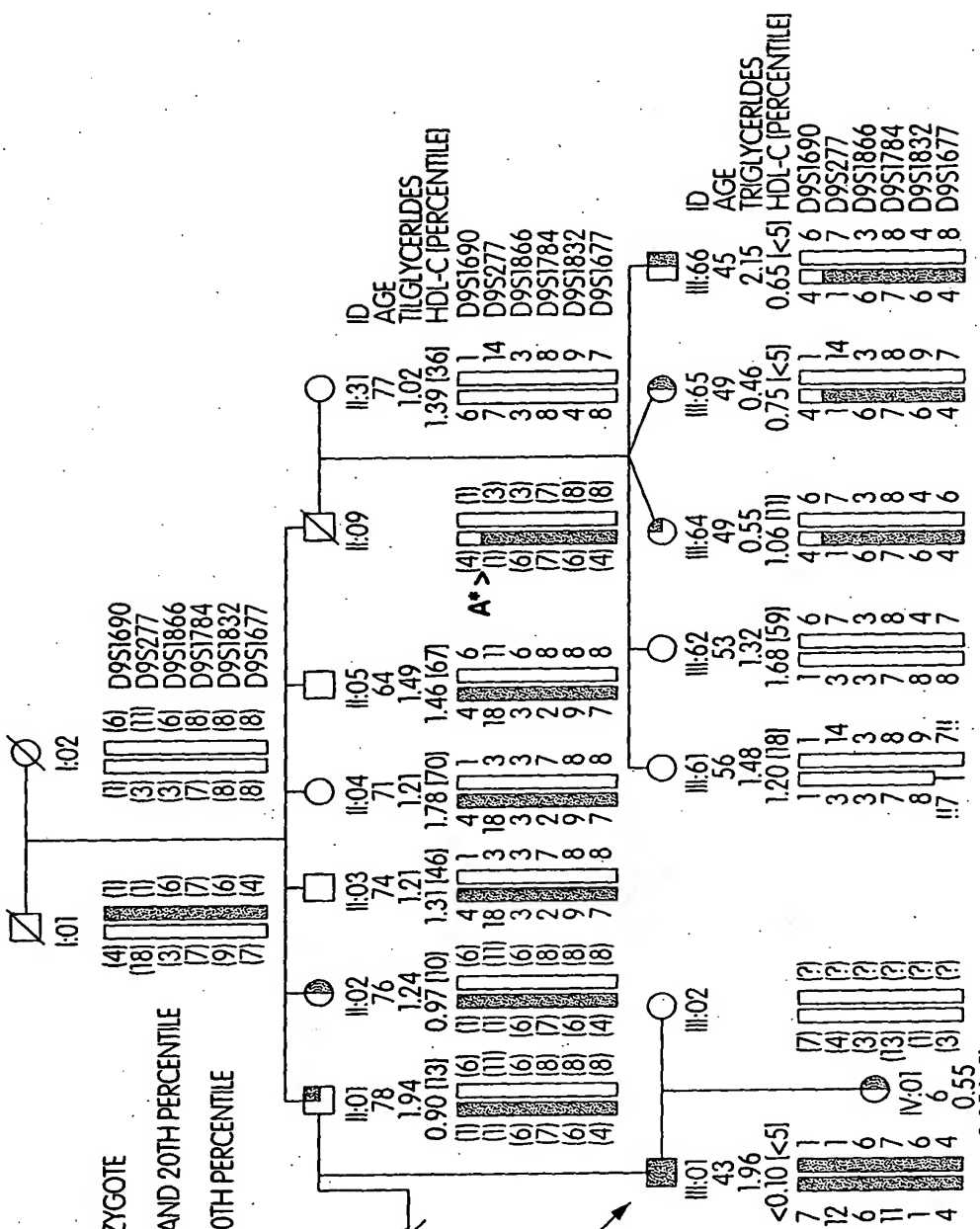
UNAFFECTED
COMPOUND HETEROZYGOTE
HDL-C BETWEEN 11TH AND 20TH PERCENTILE
HDL-C AT OR BELOW 10TH PERCENTILE
I:01
I:02
II:01
II:02
II:03
II:04
II:05
II:09
II:31
III:01
III:02
III:61
III:62
III:64
III:65
III:66
IV:01
ID
AGE
TRIGLYCERIDES
HDL-C (PERCENTILE)
D9S1690
D9S277
D9S1866
D9S1784
D9S1832
D9S1677
A*>

Figure 1A - (2)

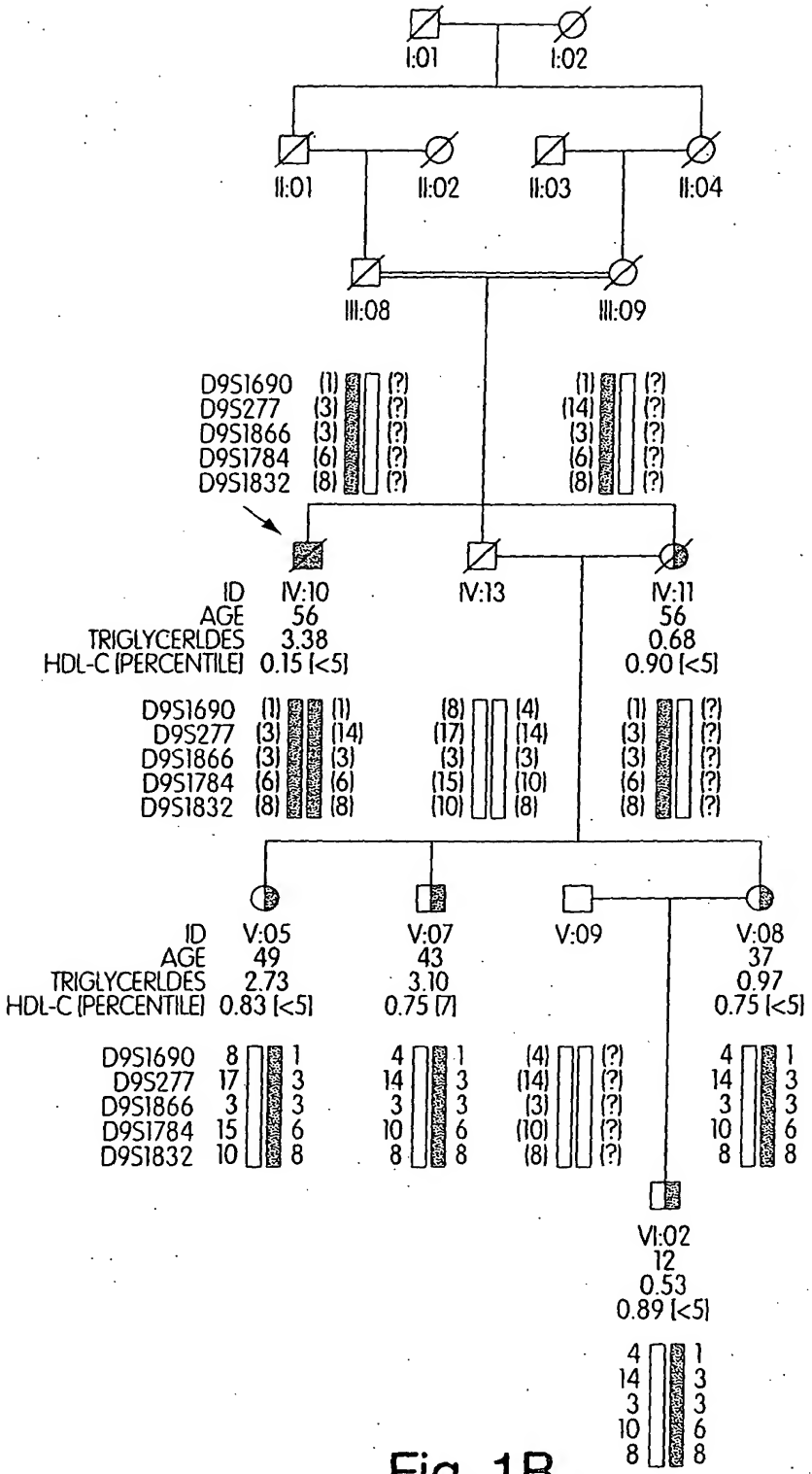
I:01
I:02
II:01
II:02
II:03
II:04
III:08
III:09
D9S1690 (1) (?)
D9S277 (3) (?)
D9S1866 (3) (?)
D9S1784 (6) (?)
D9S1832 (8) (?)
(1) (?)
(14) (?)
(3) (?)
(6) (?)
(8) (?)
ID IV:10
AGE 56
TRIGLYCERLDES 3.38
HDL-C (PERCENTILE) 0.15 (<5)
IV:13
IV:11
56
0.68
0.90 (<5)
D9S1690 (1) (1)
D9S277 (3) (14)
D9S1866 (3) (3)
D9S1784 (6) (6)
D9S1832 (8) (8)
(8) (4)
(17) (14)
(3) (3)
(15) (10)
(10) (8)
(1) (?)
(3) (?)
(3) (?)
(6) (?)
(8) (?)
ID V:05
AGE 49
TRIGLYCERLDES 2.73
HDL-C (PERCENTILE) 0.83 (<5)
V:07
43
3.10
0.75 (7)
V:09
V:08
37
0.97
0.75 (<5)
D9S1690 8 1
D9S277 17 3
D9S1866 3 3
D9S1784 15 6
D9S1832 10 8
4 1
14 3
3 3
10 6
8 8
(4) (?)
(14) (?)
(3) (?)
(10) (?)
(8) (?)
4 1
14 3
3 3
10 6
8 8
VI:02
12
0.53
0.89 (<5)
4 1
14 3
3 3
10 6
8 8

Fig. 1B

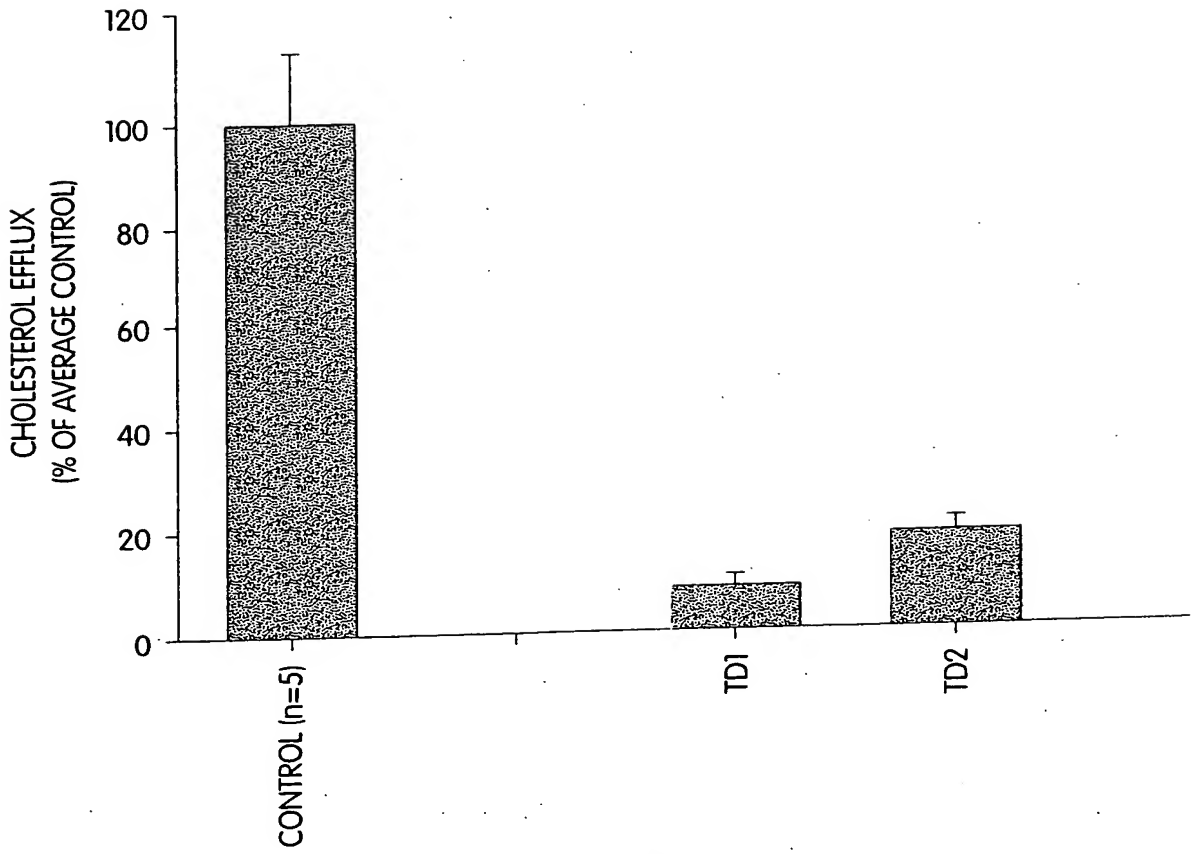
CHOLESTEROL EFFLUX
(% OF AVERAGE CONTROL)
120
100
80
60
40
20
0
CONTROL (n=5)
TD1
TD2

Fig. 1C

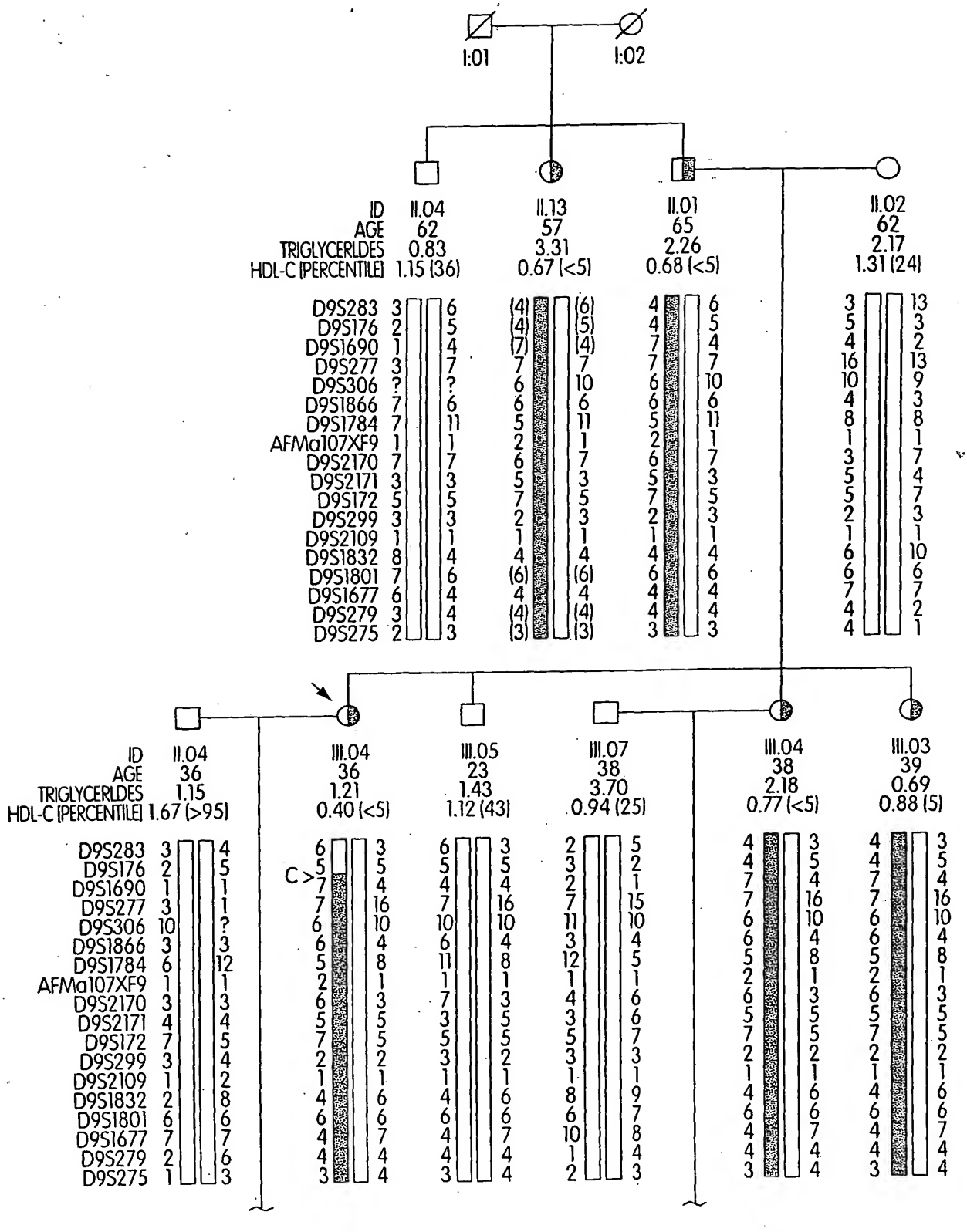
I:01
I:02
ID II.04 II.13 II.01 II.02
AGE 62 57 65 62
TRIGLYCERLDES 0.83 3.31 2.26 2.17
HDL-C (PERCENTILE) 1.15 (36) 0.67 (<5) 0.68 (<5) 1.31 (24)
D9S283 3 6 (4) (6) 4 6 3 13
D9S176 2 5 (4) (5) 4 5 5 3
D9S1690 1 4 (7) (4) 7 4 4 2
D9S277 3 7 7 7 7 7 16 13
D9S306 ? ? 6 10 6 10 10 9
D9S1866 7 6 6 6 6 6 4 3
D9S1784 7 11 5 11 5 11 8 8
AFMa107XF9 1 1 2 1 2 1 1 1
D9S2170 7 7 6 7 6 7 3 7
D9S2171 3 3 5 3 5 3 5 4
D9S172 5 5 7 5 7 5 5 7
D9S299 3 3 2 3 2 3 2 3
D9S2109 1 1 1 1 4 1 1 1
D9S1832 8 4 4 4 4 4 6 10
D9S1801 7 6 (6) (6) 6 6 6 6
D9S1677 6 4 4 4 4 4 7 7
D9S279 3 4 (4) (4) 4 4 4 2
D9S275 2 3 (3) (3) 3 3 4 1
ID II.04 III.04 III.05 III.07 III.04 III.03
AGE 36 36 23 38 38 39
TRIGLYCERLDES 1.15 1.21 1.43 3.70 2.18 0.69
HDL-C (PERCENTILE) 1.67 (>95) 0.40 (<5) 1.12 (43) 0.94 (25) 0.77 (<5) 0.88 (5)
D9S283 3 4 6 3 6 3 2 5 4 3 4 3
D9S176 2 5 C> 5 5 5 5 3 2 4 5 4 5
D9S1690 1 1 7 4 4 4 2 1 7 4 7 4
D9S277 3 1 7 16 7 16 7 15 7 16 7 16
D9S306 10 ? 6 10 10 10 11 10 6 10 6 10
D9S1866 3 3 6 4 6 4 3 4 6 4 6 4
D9S1784 6 12 5 8 11 8 12 5 5 8 5 8
AFMa107XF9 1 1 2 1 1 1 1 6 2 1 2 1
D9S2170 3 3 6 3 7 3 4 6 6 3 6 3
D9S2171 4 4 5 5 3 5 3 6 5 5 5 5
D9S172 7 5 7 5 5 5 5 7 7 2 7 2
D9S299 3 4 2 2 3 2 1 3 2 1 2 1
D9S2109 1 2 1 1 1 6 8 1 1 4 1 6
D9S1832 2 8 4 6 4 6 6 4 6 4 6 4
D9S1801 6 6 6 7 4 7 10 4 4 4 4 4
D9S1677 7 7 4 4 4 4 1 3 4 4 4 4
D9S279 2 6 4 4 4 4 2 4 3 4 3 4
D9S275 1 3 3 4 3 4 3 3 2 3 3 4

Figure 2A – (1)

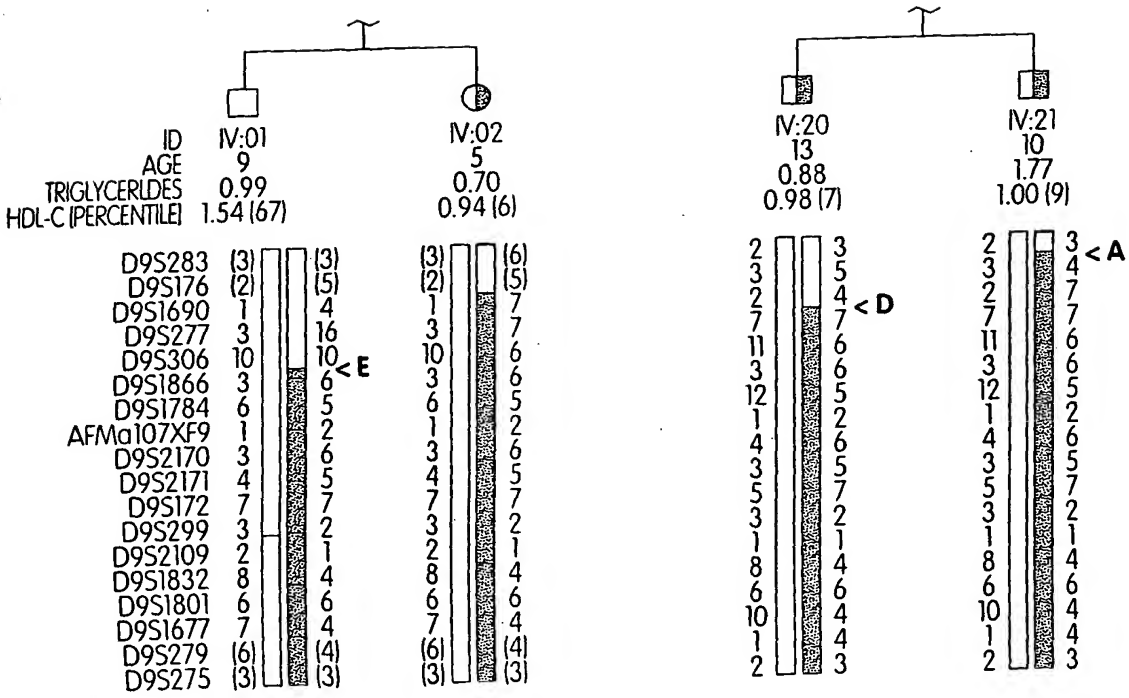
ID
AGE
TRIGLYCERIDES
HDL-C (PERCENTILE)
IV:01
9
0.99
1.54 (67)
IV:02
5
0.70
0.94 (6)
IV:20
13
0.88
0.98 (7)
IV:21
10
1.77
1.00 (9)
D9S283
D9S176
D9S1690
D9S277
D9S306
D9S1866
D9S1784
AFMa107XF9
D9S2170
D9S2171
D9S172
D9S299
D9S2109
D9S1832
D9S1801
D9S1677
D9S279
D9S275
< E
< D
< A

Figure 2A – (2)

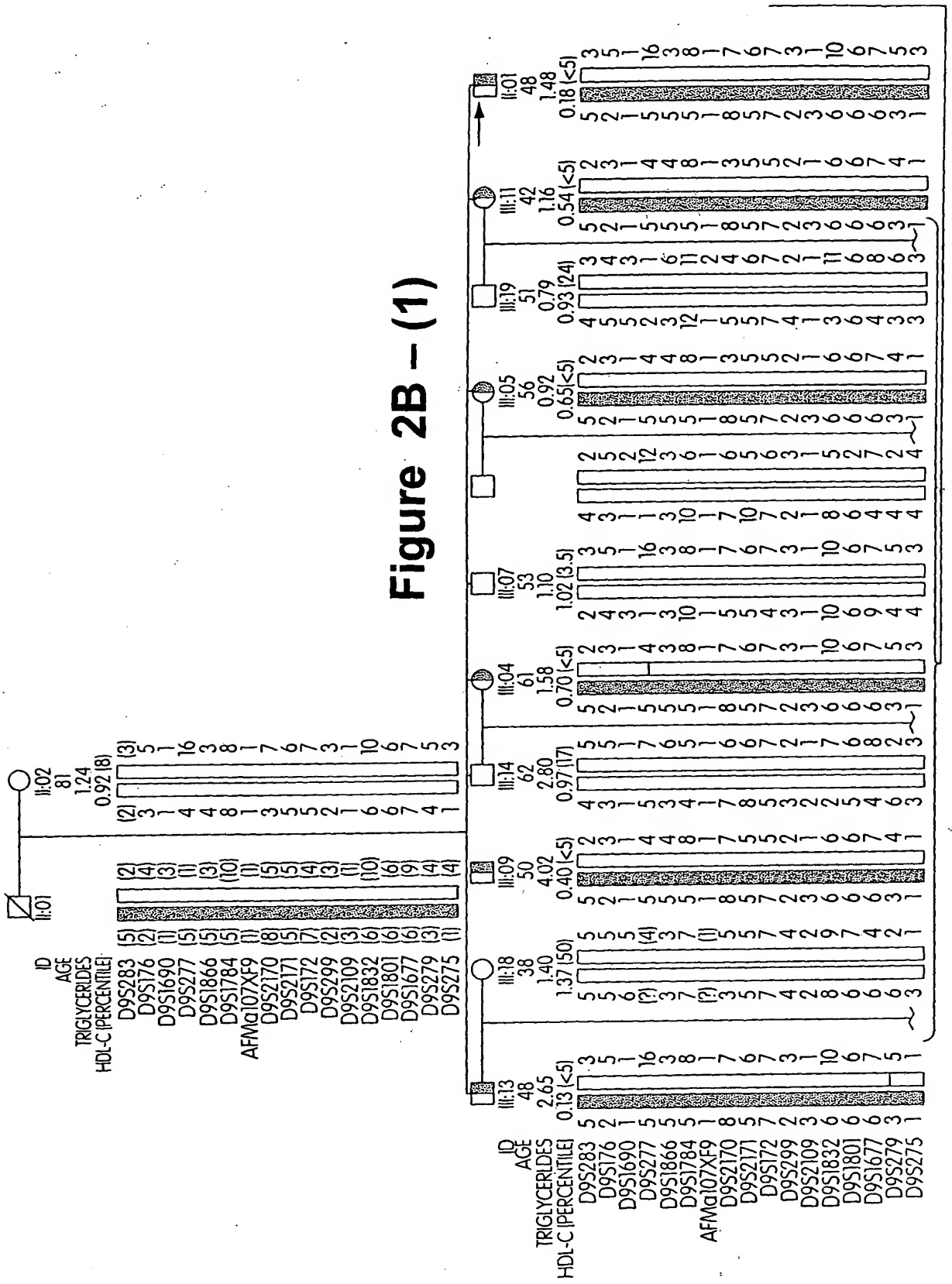
ID
AGE
TRIGLYCERIDES
HDL-C (PERCENTILE)
D9S283
D9S176
D9S1690
D9S277
D9S1866
D9S1784
AFMa107XF9
D9S2170
D9S2171
D9S172
D9S299
D9S2109
D9S1832
D9S1801
D9S1677
D9S279
D9S275
I:01
II:02
81
1.24
0.92 (8)
III:13
48
2.65
0.13 (<5)
III:18
38
1.40
1.37 (50)
III:09
50
4.02
0.40 (<5)
III:14
62
2.80
0.97 (17)
III:04
61
1.58
0.70 (<5)
III:07
53
1.10
1.02 (3.5)
III:05
56
0.92
0.65 (<5)
III:19
51
0.79
0.93 (24)
III:11
42
1.16
0.54 (<5)
II:01
48
1.48
0.18 (<5)

Figure 2B – (1)

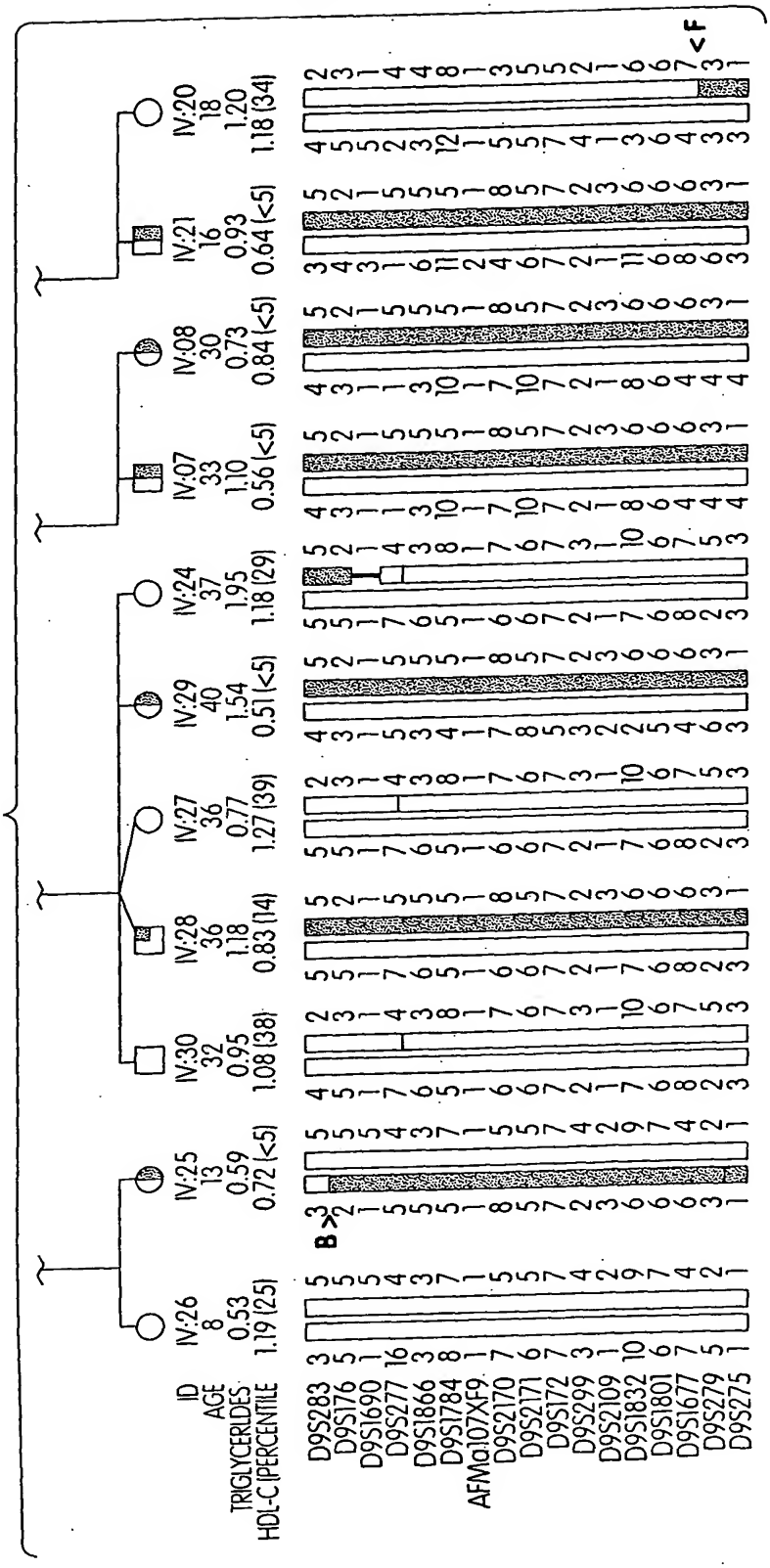
ID
AGE
TRIGLYCERIDES
HDL-C (PERCENTILE
IV:26
8
0.53
1.19 (25)
IV:25
13
0.59
0.72 (<5)
IV:30
32
0.95
1.08 (38)
IV:28
36
1.18
0.83 (14)
IV:27
36
0.77
1.27 (39)
IV:29
40
1.54
0.51 (<5)
IV:24
37
1.95
1.18 (29)
IV:07
33
1.10
0.56 (<5)
IV:08
30
0.73
0.84 (<5)
IV:21
16
0.93
0.64 (<5)
IV:20
18
1.20
1.18 (34)
D9S283
D9S176
D9S1690
D9S277
D9S1866
D9S1784
AFMa107XF9
D9S2170
D9S2171
D9S172
D9S299
D9S2109
D9S1832
D9S1801
D9S1677
D9S279
D9S275
B >
< F

Figure 2B – (2)

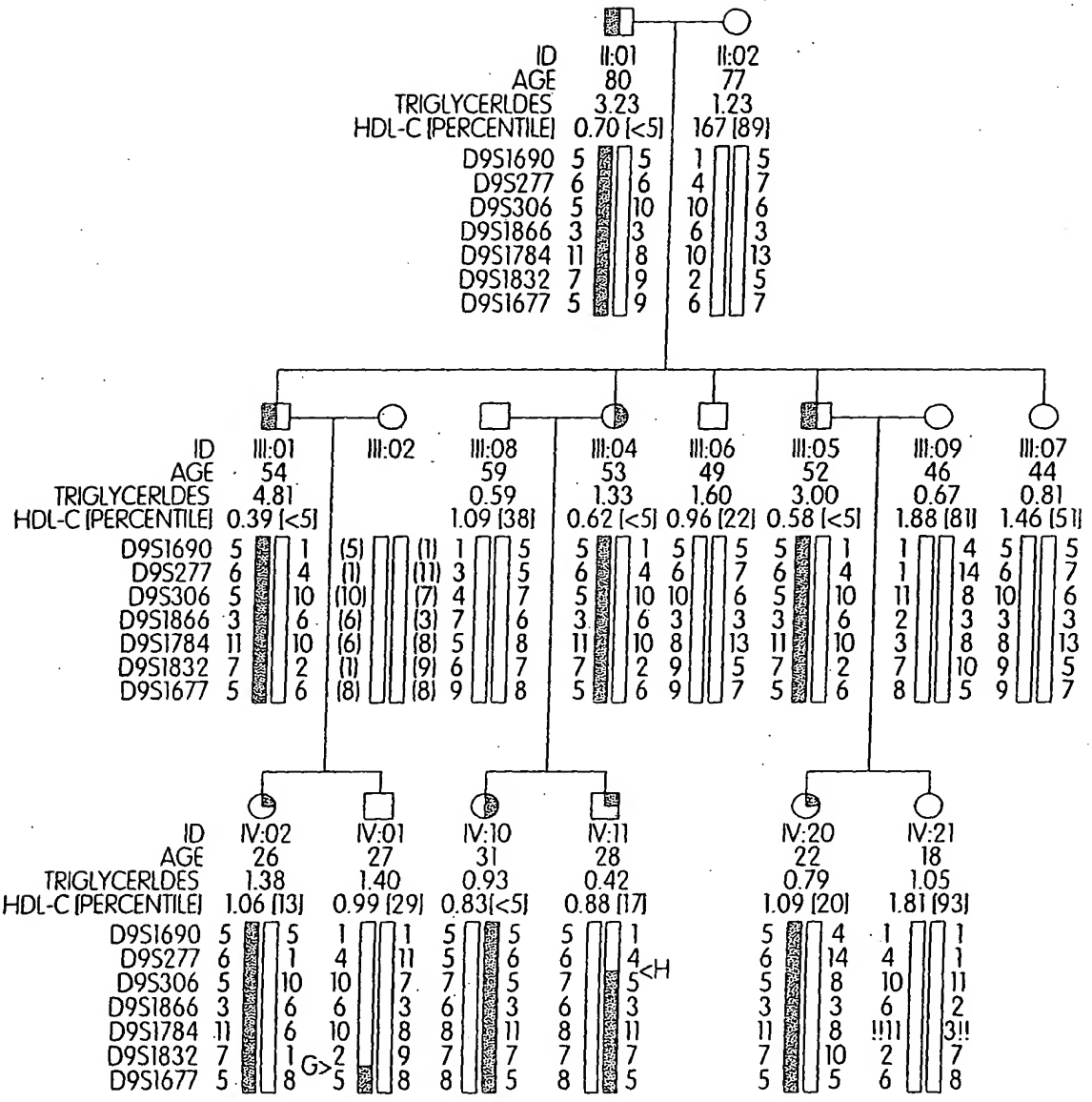
ID II:01 II:02
AGE 80 77
TRIGLYCERLDES 3.23 1.23
HDL-C (PERCENTILE) 0.70 (<5) 167 (89)
D9S1690 5 5 1 5
D9S277 6 6 4 7
D9S306 5 10 10 6
D9S1866 3 3 6 3
D9S1784 11 8 10 13
D9S1832 7 9 2 5
D9S1677 5 9 6 7
ID III:01 III:02 III:08 III:04 III:06 III:05 III:09 III:07
AGE 54 59 53 49 52 46 44
TRIGLYCERLDES 4.81 0.59 1.33 1.60 3.00 0.67 0.81
HDL-C (PERCENTILE) 0.39 (<5) 1.09 (38) 0.62 (<5) 0.96 (22) 0.58 (<5) 1.88 (81) 1.46 (51)
D9S1690 5 1 (5) (1) 1 5 5 1 5 5 1 4 5 5
D9S277 6 4 (1) (11) 3 5 6 4 6 7 6 4 1 14 6 7
D9S306 5 10 (10) (7) 4 7 5 10 10 6 5 10 11 8 10 6
D9S1866 3 6 (6) (3) 7 6 3 6 3 3 3 6 2 3 3 3
D9S1784 11 10 (6) (8) 5 8 11 10 8 13 11 10 3 8 8 13
D9S1832 7 2 (1) (9) 6 7 7 2 9 5 7 2 7 10 9 5
D9S1677 5 6 (8) (8) 9 8 5 6 9 7 5 6 8 5 9 7
ID IV:02 IV:01 IV:10 IV:11 IV:20 IV:21
AGE 26 27 31 28 22 18
TRIGLYCERLDES 1.38 1.40 0.93 0.42 0.79 1.05
HDL-C (PERCENTILE) 1.06 (13) 0.99 (29) 0.83(<5) 0.88 (17) 1.09 (20) 1.81 (93)
D9S1690 5 5 1 1 5 5 5 1 5 4 1 1
D9S277 6 1 4 11 5 6 6 4 6 14 4 1
D9S306 5 10 10 7 7 5 7 5<H 5 8 10 11
D9S1866 3 6 6 3 6 3 6 3 3 3 6 2
D9S1784 11 6 10 8 8 11 8 11 11 8 !!!! 3!!
D9S1832 7 1 2 9 7 7 7 7 7 10 2 7
D9S1677 5 8 G> 5 8 8 5 8 5 5 5 6 8

Fig. 2C

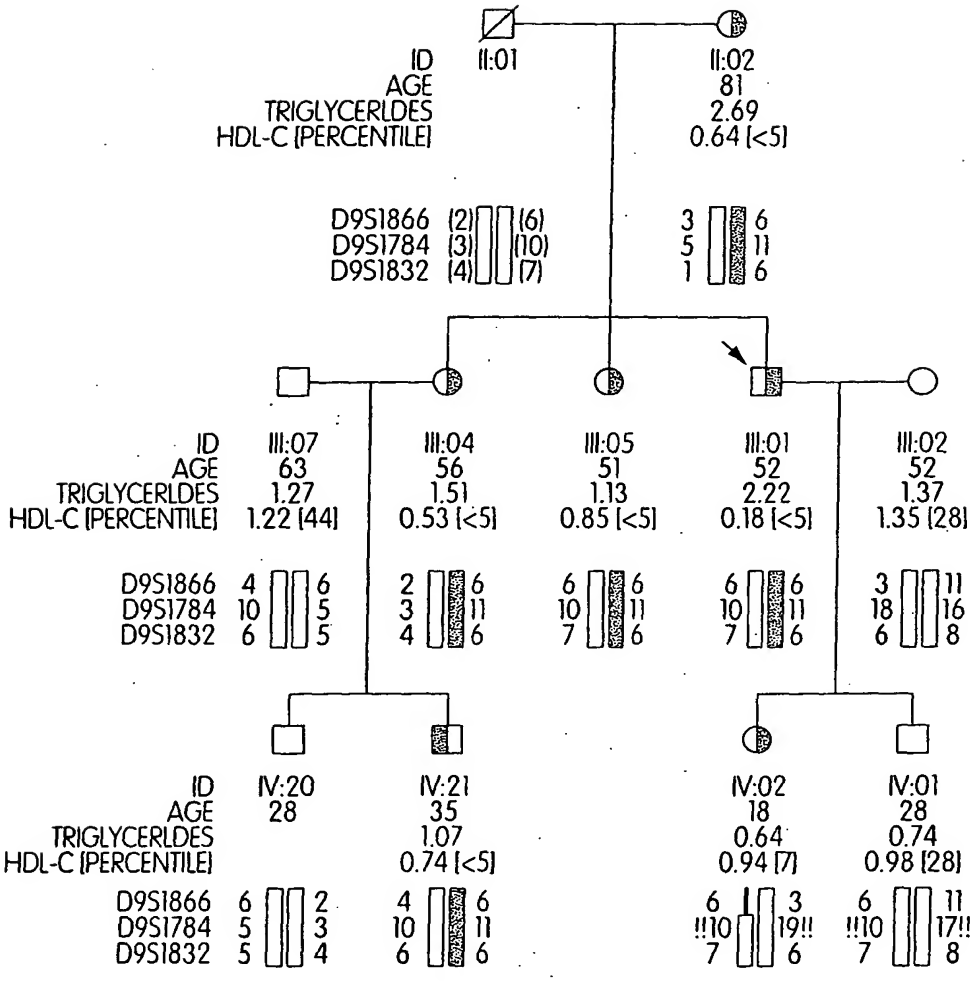
ID II:01 II:02
AGE 81
TRIGLYCERLDES 2.69
HDL-C (PERCENTILE) 0.64 (<5)
D9S1866 (2) (6) 3 6
D9S1784 (3) (10) 5 11
D9S1832 (4) (7) 1 6
ID III:07 III:04 III:05 III:01 III:02
AGE 63 56 51 52 52
TRIGLYCERLDES 1.27 1.51 1.13 2.22 1.37
HDL-C (PERCENTILE) 1.22 (44) 0.53 (<5) 0.85 (<5) 0.18 (<5) 1.35 (28)
D9S1866 4 6 2 6 6 6 6 6 3 11
D9S1784 10 5 3 11 10 11 10 11 18 16
D9S1832 6 5 4 6 7 6 7 6 6 8
ID IV:20 IV:21 IV:02 IV:01
AGE 28 35 18 28
TRIGLYCERLDES 1.07 0.64 0.74
HDL-C (PERCENTILE) 0.74 (<5) 0.94 (7) 0.98 (28)
D9S1866 6 2 4 6 6 3 6 11
D9S1784 5 3 10 11 !!10 19!! !!10 17!!
D9S1832 5 4 6 6 7 6 7 8

Fig. 2D

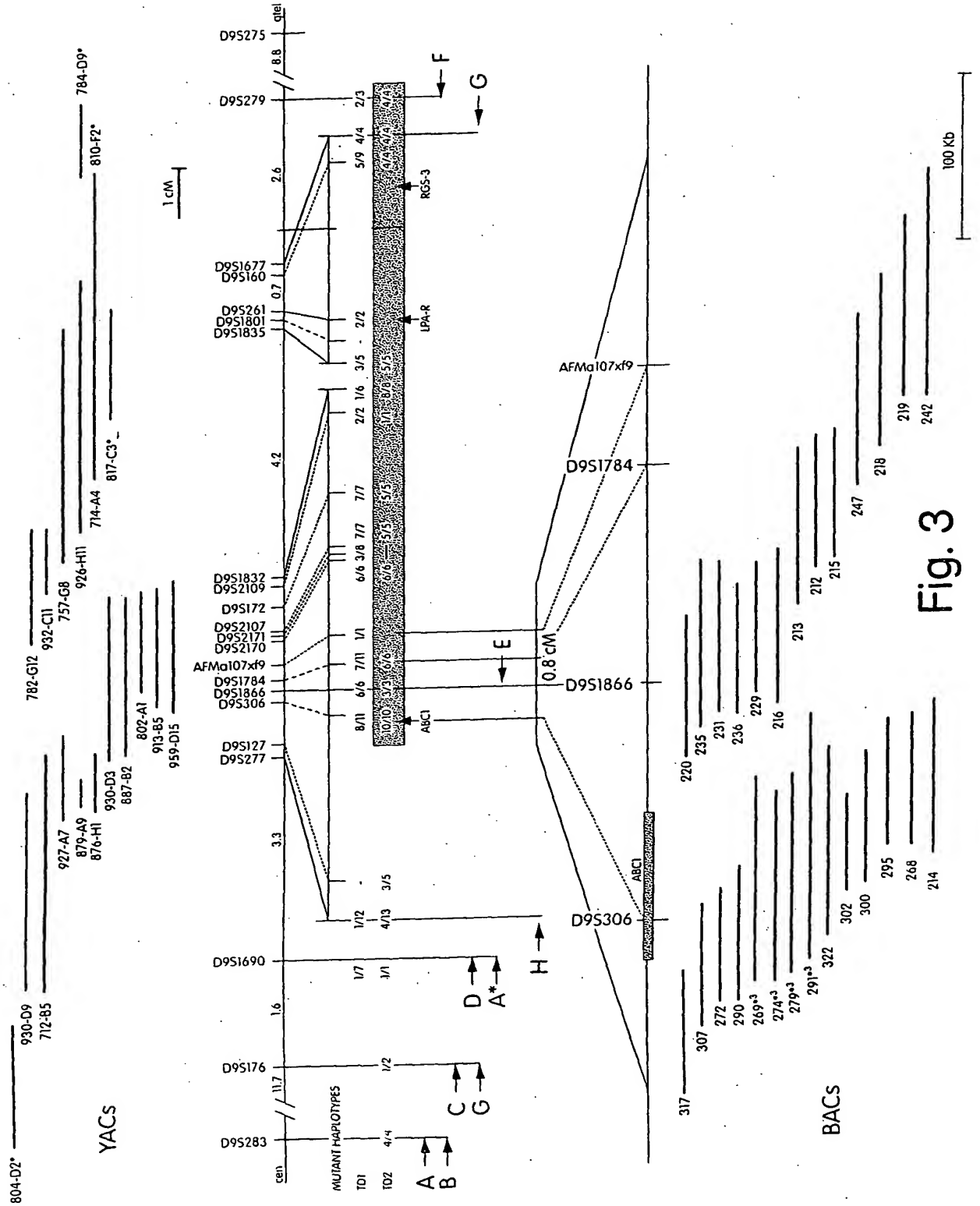
YACs
BACs
MUTANT HAPLOTYPES
cen
qtel
1 cM
0.8 cM
100 Kb
D9S275
D9S279
D9S1677
D9S160
D9S261
D9S1801
D9S1835
D9S1832
D9S2109
D9S172
D9S2107
D9S2171
D9S2170
AFMa107xf9
D9S1784
D9S1866
D9S306
D9S127
D9S277
D9S1690
D9S176
D9S283
RGS-3
LPA-R
ABC1
804-D2*
930-D9
712-B5
927-A7
879-A9
876-H1
930-D3
887-B2
802-A1
913-B5
959-D15
782-G12
932-C11
757-G8
926-H11
714-A4
817-C3*
810-F2*
784-D9*
A
B
C
G
D
A*
H
E
F
G

Fig. 3

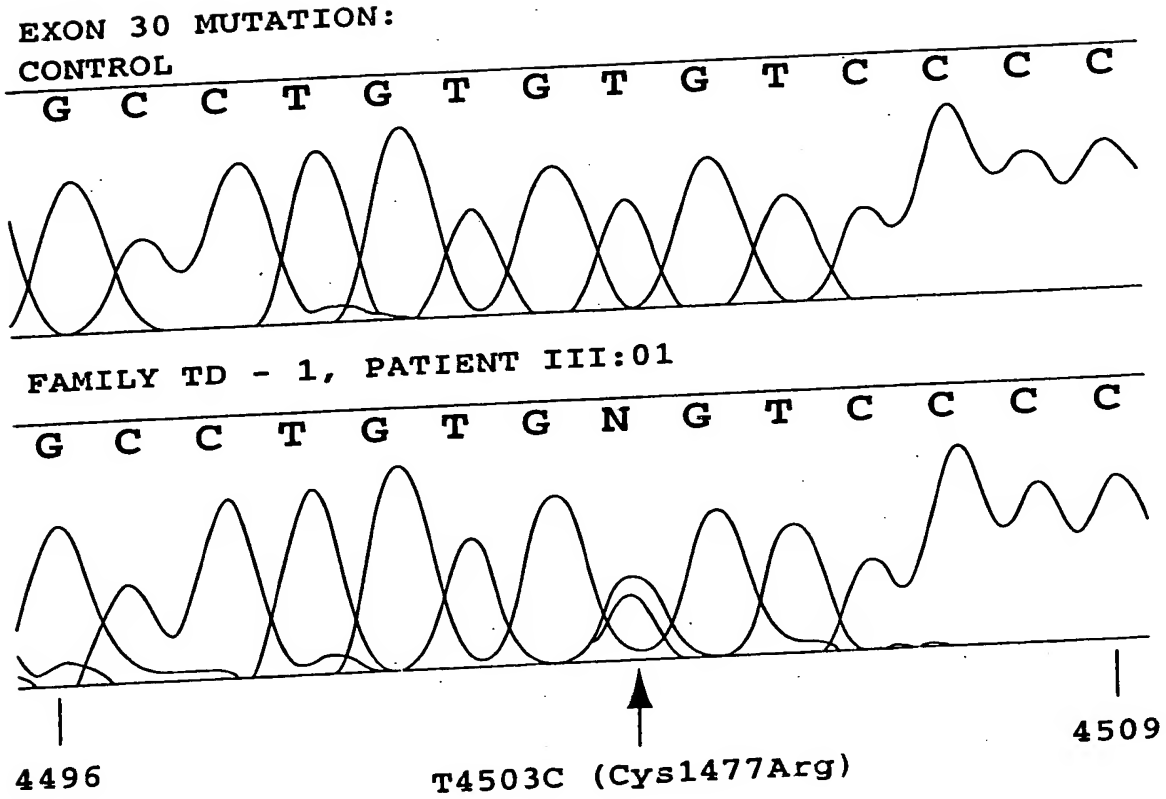
EXON 30 MUTATION:
CONTROL
G C C T G T G T G T C C C C
FAMILY TD - 1, PATIENT III:01
G C C T G T G N G T C C C C
4496
T4503C (Cys1477Arg)
4509

Fig. 4A

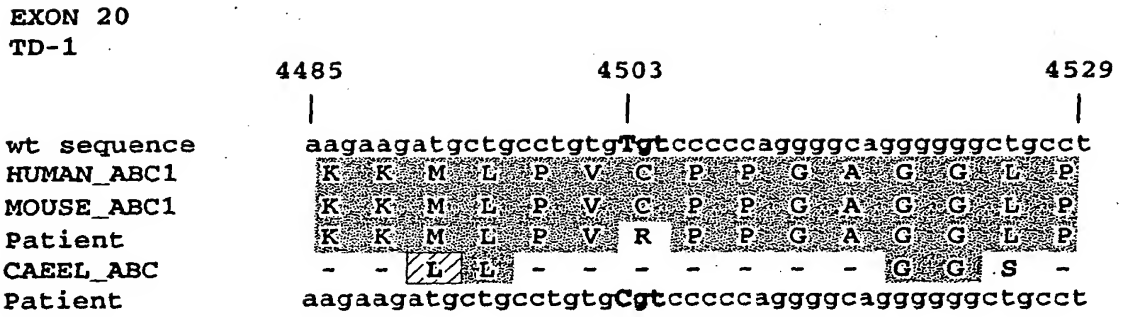
EXON 20
TD-1
4485
4503
4529
wt sequence
aagaagatgctgcctgtgTgtccccccaggggcaggggggctgcct
HUMAN_ABC1
K K M L P V C P P G A G G L P
MOUSE_ABC1
K K M L P V C P P G A G G L P
Patient
K K M L P V R P P G A G G L P
CAEEL_ABC
- - L L - - - - - - - - G G S -
Patient
aagaagatgctgcctgtgCgtccccccaggggcaggggggctgcct

Fig. 4B

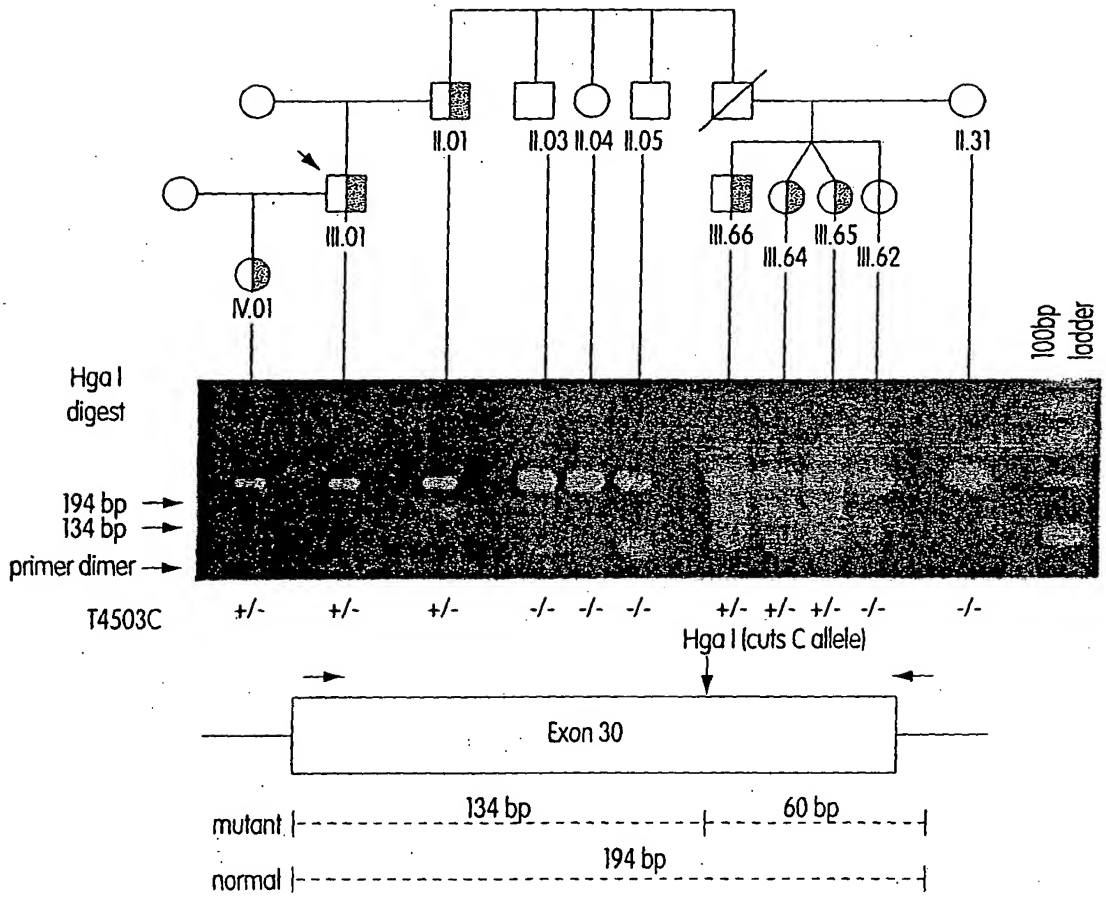
II.01
II.03
II.04
II.05
II.31
III.01
III.66
III.64
III.65
III.62
IV.01
100bp ladder
Hga I digest
194 bp
134 bp
primer dimer
T4503C
+/- +/- +/- -/- -/- -/- +/- +/- +/- -/- -/-
Hga I (cuts C allele)
Exon 30
mutant
134 bp
60 bp
normal
194 bp

Fig. 4C

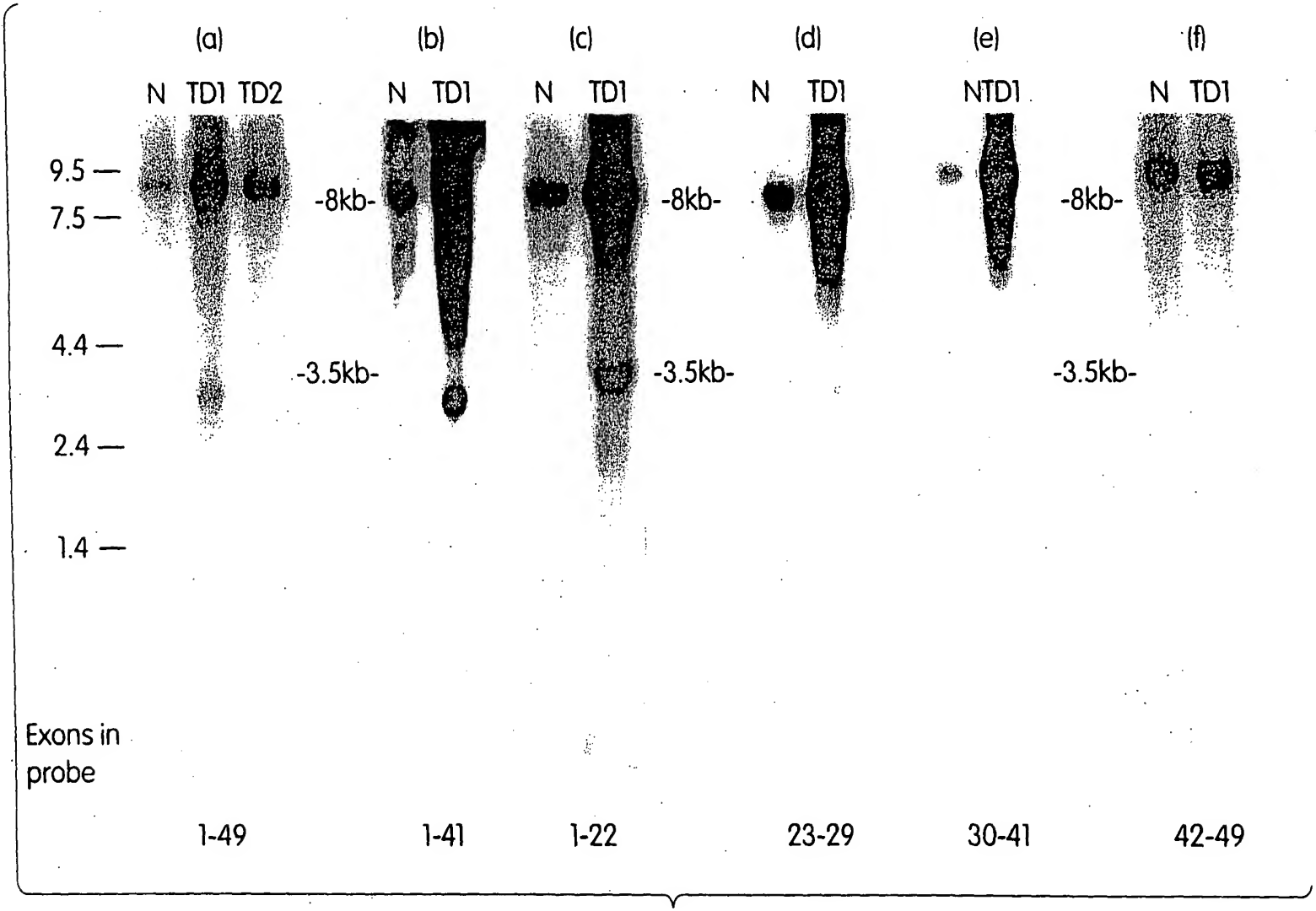
(a)
(b)
(c)
(d)
(e)
(f)
N TD1 TD2
N TD1
N TD1
N TD1
NTD1
N TD1
9.5
7.5
4.4
2.4
1.4
-8kb-
-3.5kb-
-8kb-
-3.5kb-
-8kb-
-3.5kb-
Exons in probe
1-49
1-41
1-22
23-29
30-41
42-49

Fig. 4D

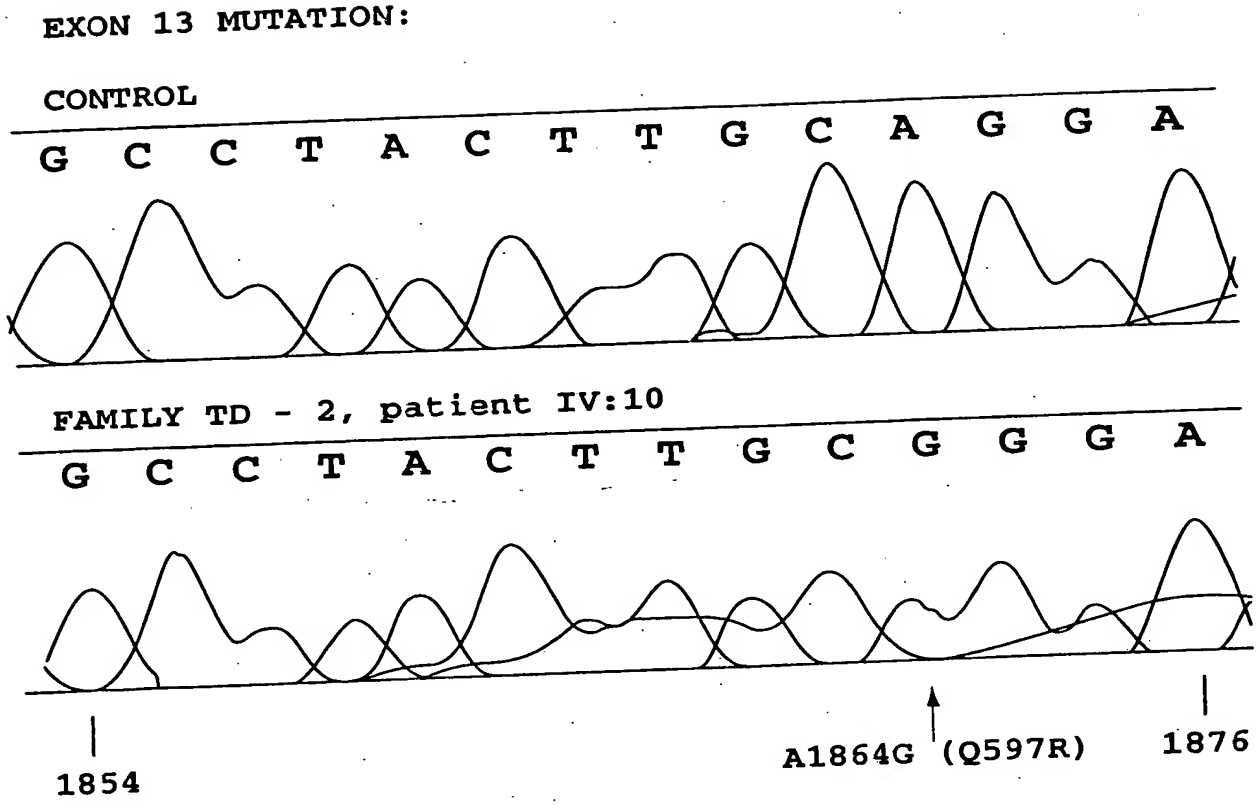
EXON 13 MUTATION:
CONTROL
G C C T A C T T G C A G G A
FAMILY TD - 2, patient IV:10
G C C T A C T T G C G G G A
1854
A1864G (Q597R)
1876

Fig. 5A

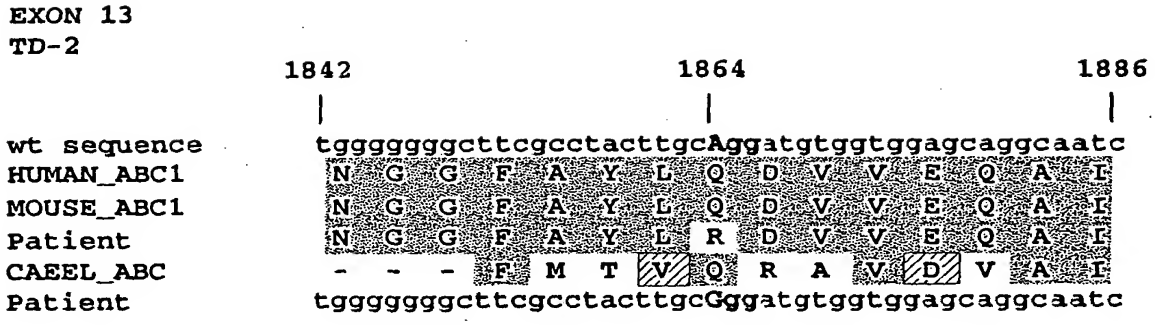
EXON 13
TD-2
1842
1864
1886
wt sequence
HUMAN_ABC1
MOUSE_ABC1
Patient
CAEEL_ABC
Patient
tgggggggcttcgcctacttgcAggatgtggtggagcaggcaatc
N G G F A Y L Q D V V E Q A I
N G G F A Y L Q D V V E Q A I
N G G F A Y L R D V V E Q A I
- - - F M T V Q R A V D V A I
tgggggggcttcgcctacttgcGggatgtggtggagcaggcaatc

Fig. 5B

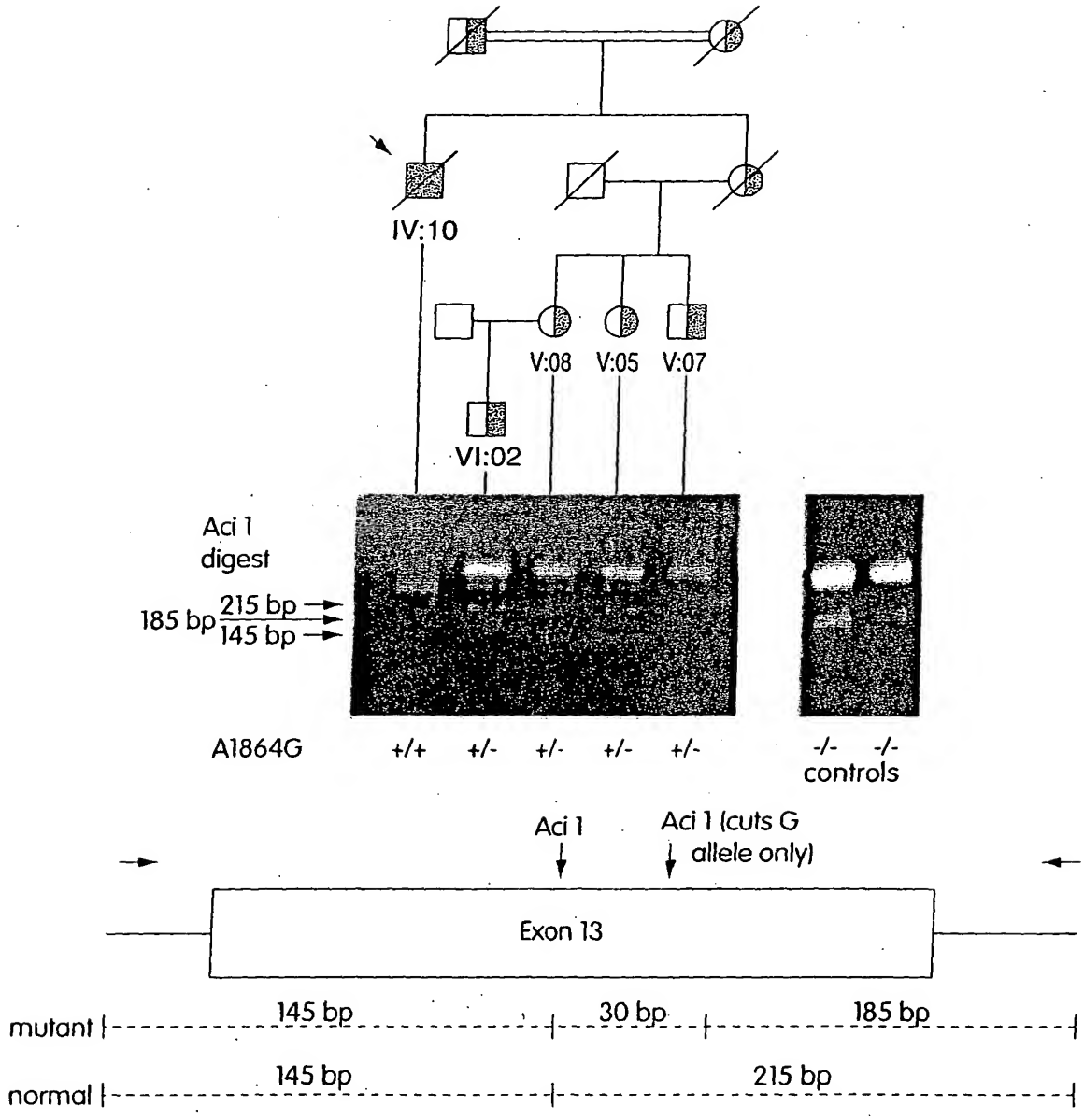
IV:10
V:08
V:05
V:07
VI:02
Aci 1
digest
185 bp
215 bp
145 bp
A1864G
+/+
+/-
+/-
+/-
+/-
-/-
-/-
controls
Aci 1
Aci 1 (cuts G
allele only)
Exon 13
mutant
145 bp
30 bp
185 bp
normal
145 bp
215 bp

Fig. 5C

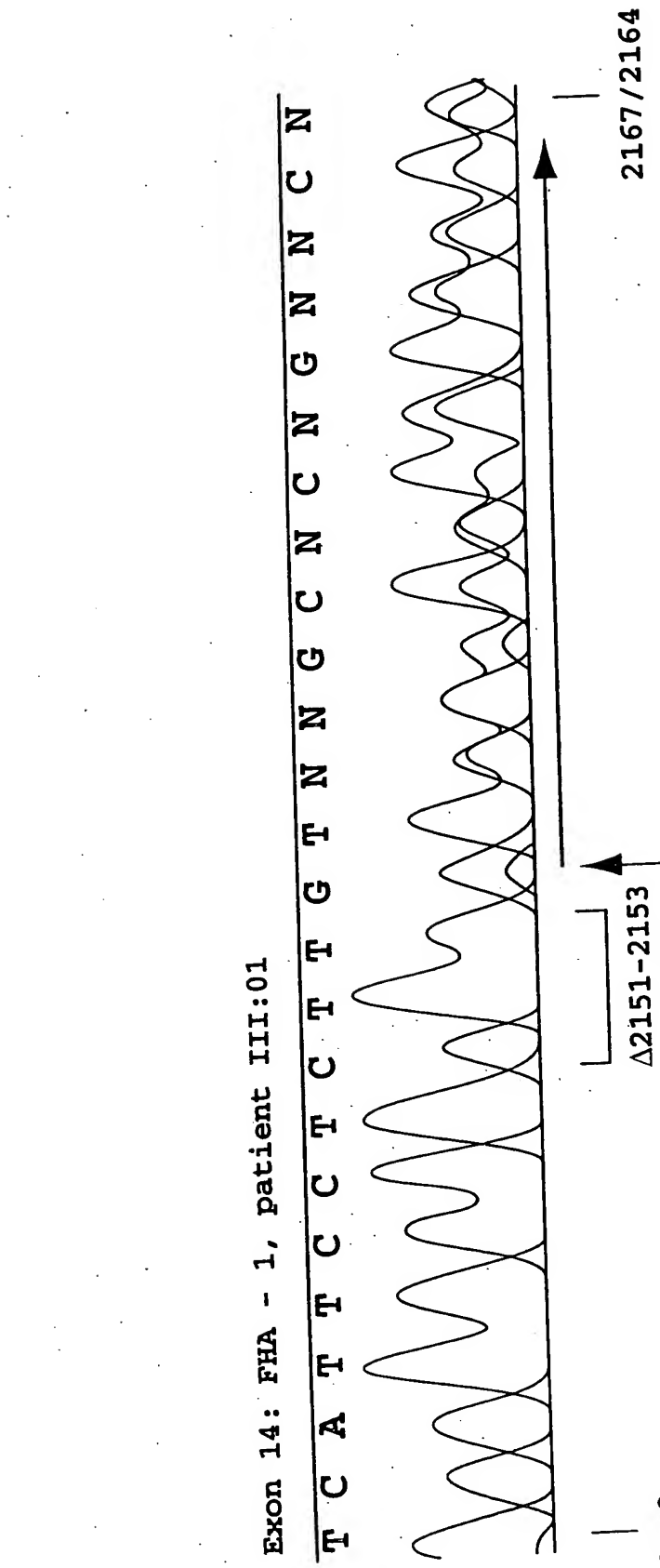
Exon 14: FHA - 1, patient III:01
T C A T T C C T C T T G T N N G G C N C N G N N N C N
2143
2154
Δ2151-2153
(ΔL693)
2167/2164

Fig. 6A

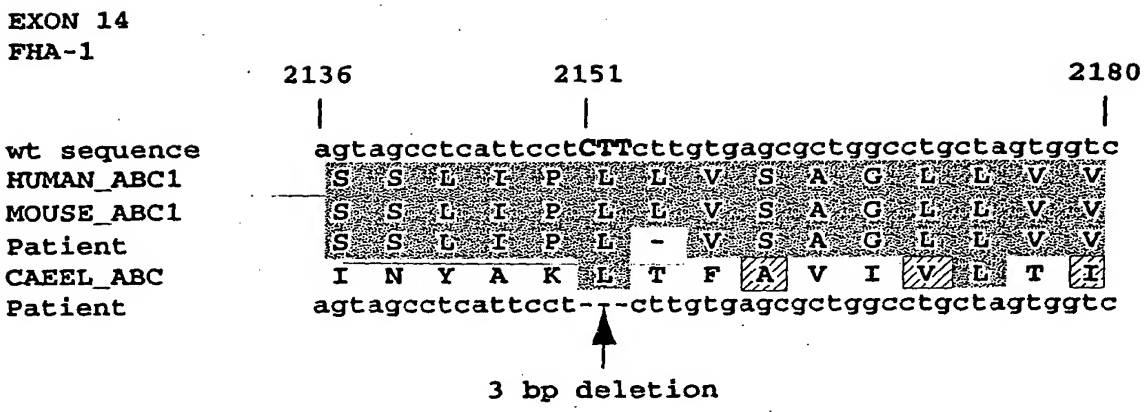
EXON 14
FHA-1
2136
2151
2180
wt sequence
HUMAN_ABC1
MOUSE_ABC1
Patient
CAEEL_ABC
Patient
agtagcctcattcctCTTcttgtgagcgctggcctgctagtggtc
S S L I P L L V S A G L L V V
S S L I P L L V S A G L L V V
S S L I P L - V S A G L L V V
I N Y A K L T F A V I V L T I
agtagcctcattcct---cttgtgagcgctggcctgctagtggtc
3 bp deletion

Fig. 6B

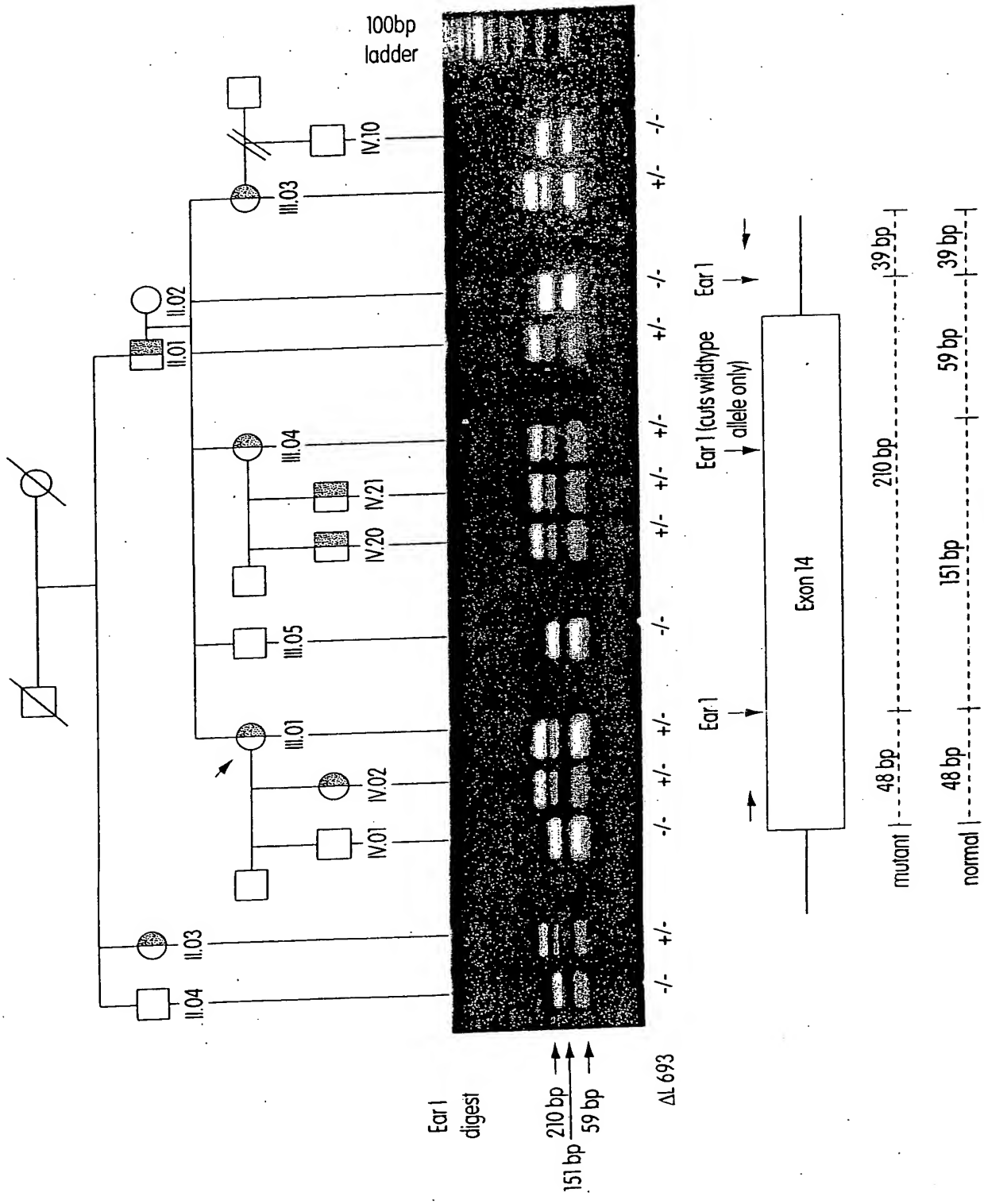
100bp ladder
II.03
II.04
II.01
II.02
III.01
III.03
III.04
III.05
IV.01
IV.02
IV.10
IV.20
IV.21
-/- +/- +/- +/- -/- +/- +/- +/- -/-
Ear I digest
151 bp
210 bp
59 bp
ΔL 693
Ear 1
Ear 1 (cuts wildtype allele only)
Exon 14
48 bp
210 bp
39 bp
mutant
48 bp
151 bp
59 bp
39 bp
normal

Fig. 6C

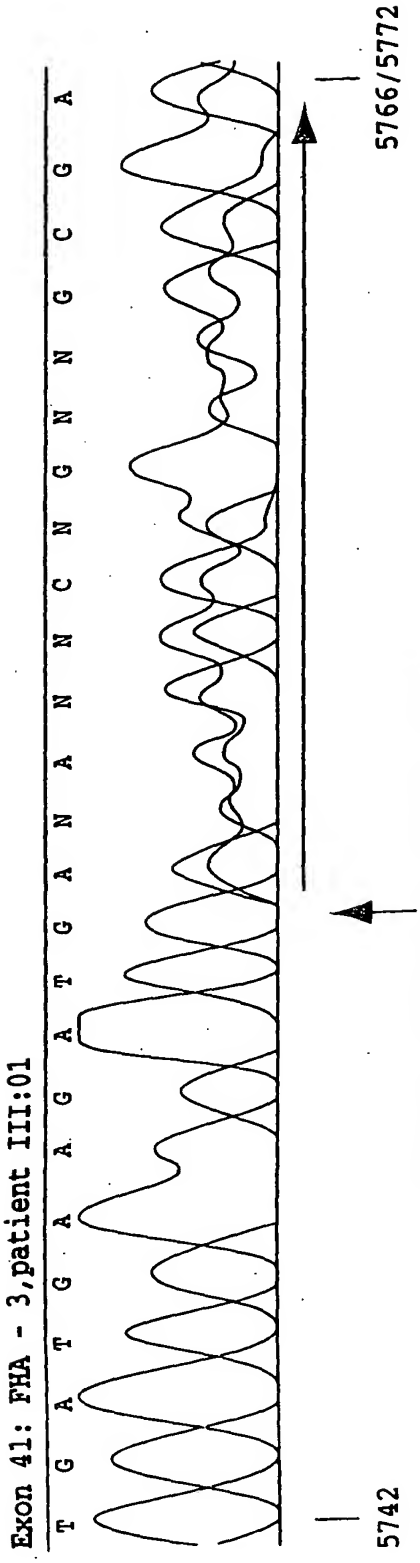
Exon 41: FHA - 3,patient III:01
T G A T G A A G A T G A N N A N N C N G N N G C G A
5742
5766/5772
Start of Δ5752-5757 (ΔE1893, D1894)

Fig. 6D

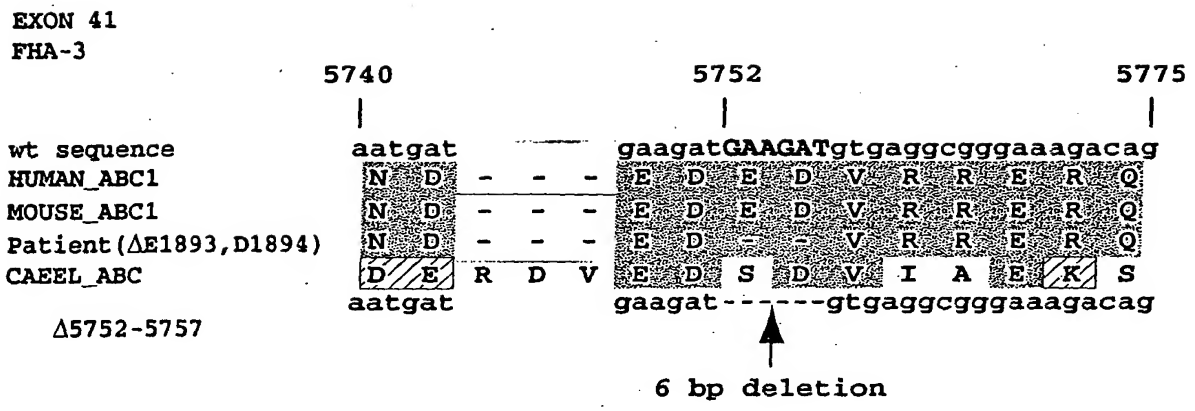
EXON 41
FHA-3
5740
5752
5775
wt sequence
HUMAN_ABC1
MOUSE_ABC1
Patient(ΔE1893,D1894)
CAEEL_ABC
aatgat
gaagatGAAGATgtgaggcgggaaagacag
N D - - - E D E D V R R E R Q
N D - - - E D E D V R R E R Q
N D - - - E D - - V R R E R Q
D E R D V E D S D V I A E K S
aatgat
gaagat------gtgaggcgggaaagacag
Δ5752-5757
6 bp deletion

Fig. 6E

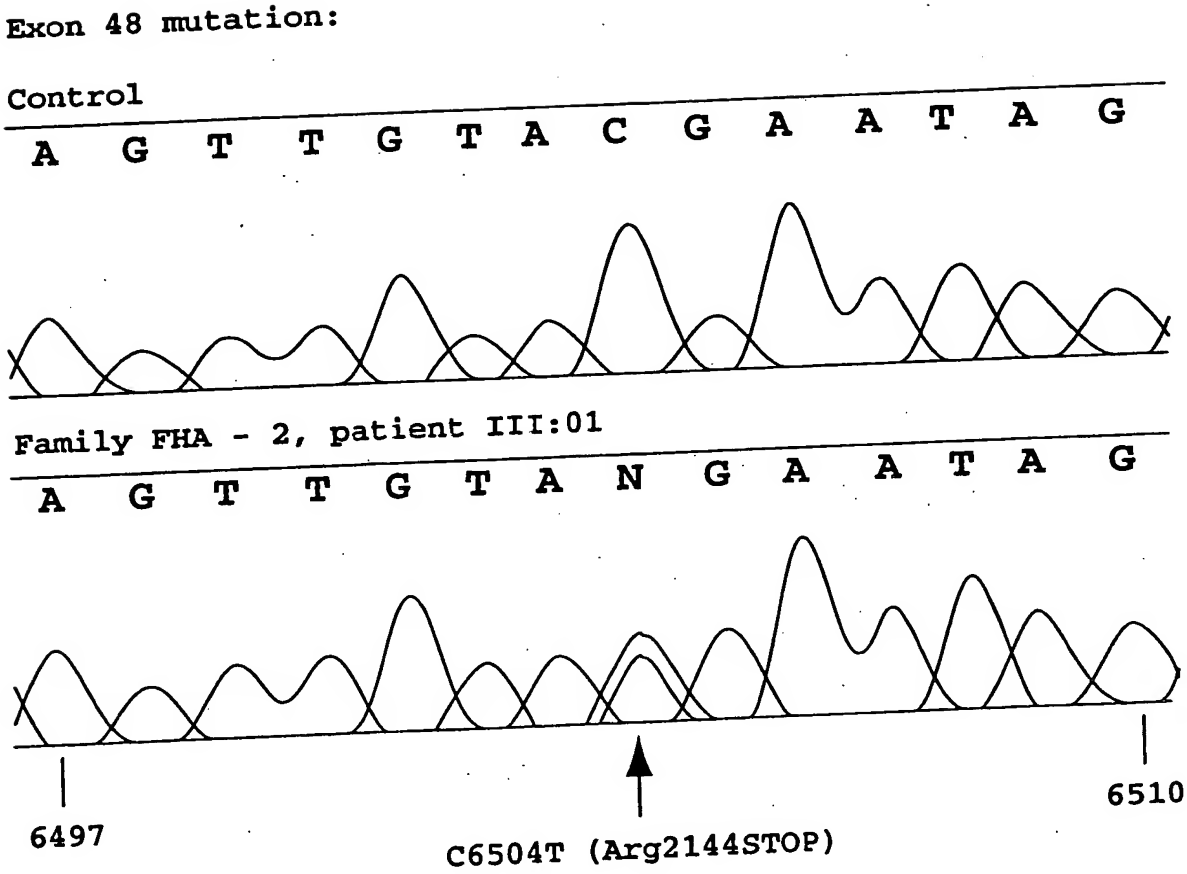
Exon 48 mutation:
Control
A G T T G T A C G A A T A G
Family FHA - 2, patient III:01
A G T T G T A N G A A T A G
6497
C6504T (Arg2144STOP)
6510

Fig. 6F

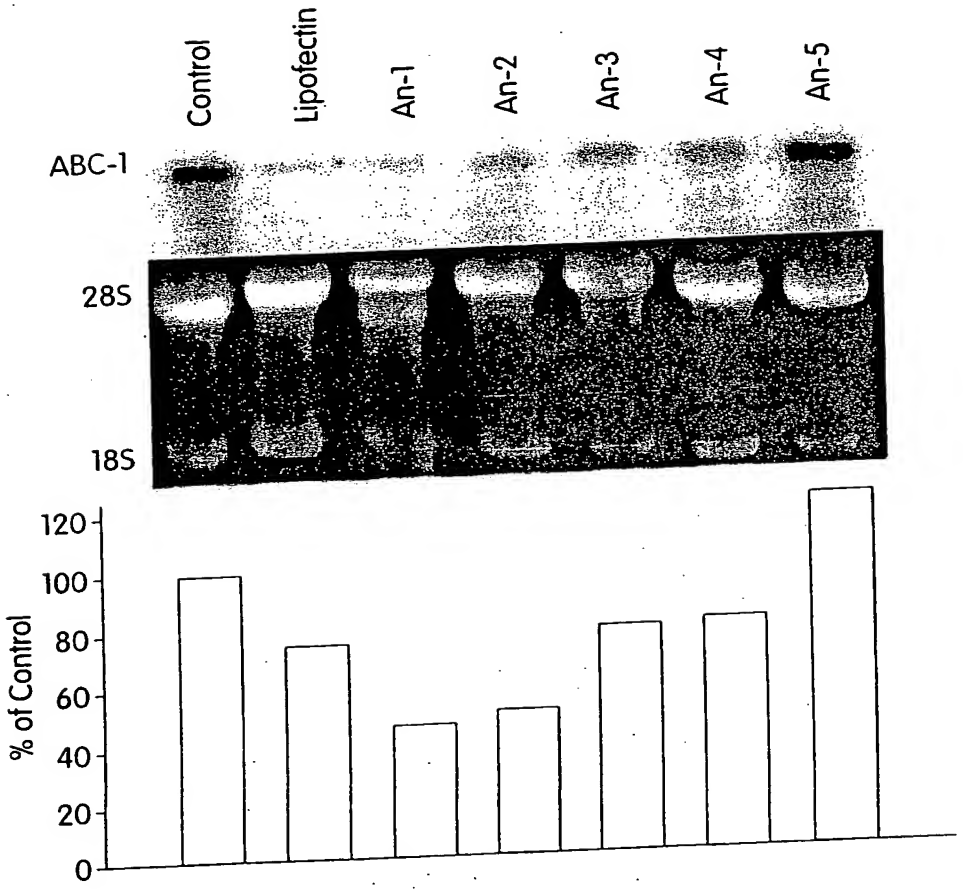
Control
Lipofectin
An-1
An-2
An-3
An-4
An-5
ABC-1
28S
18S
% of Control
120
100
80
60
40
20
0

Fig. 7A

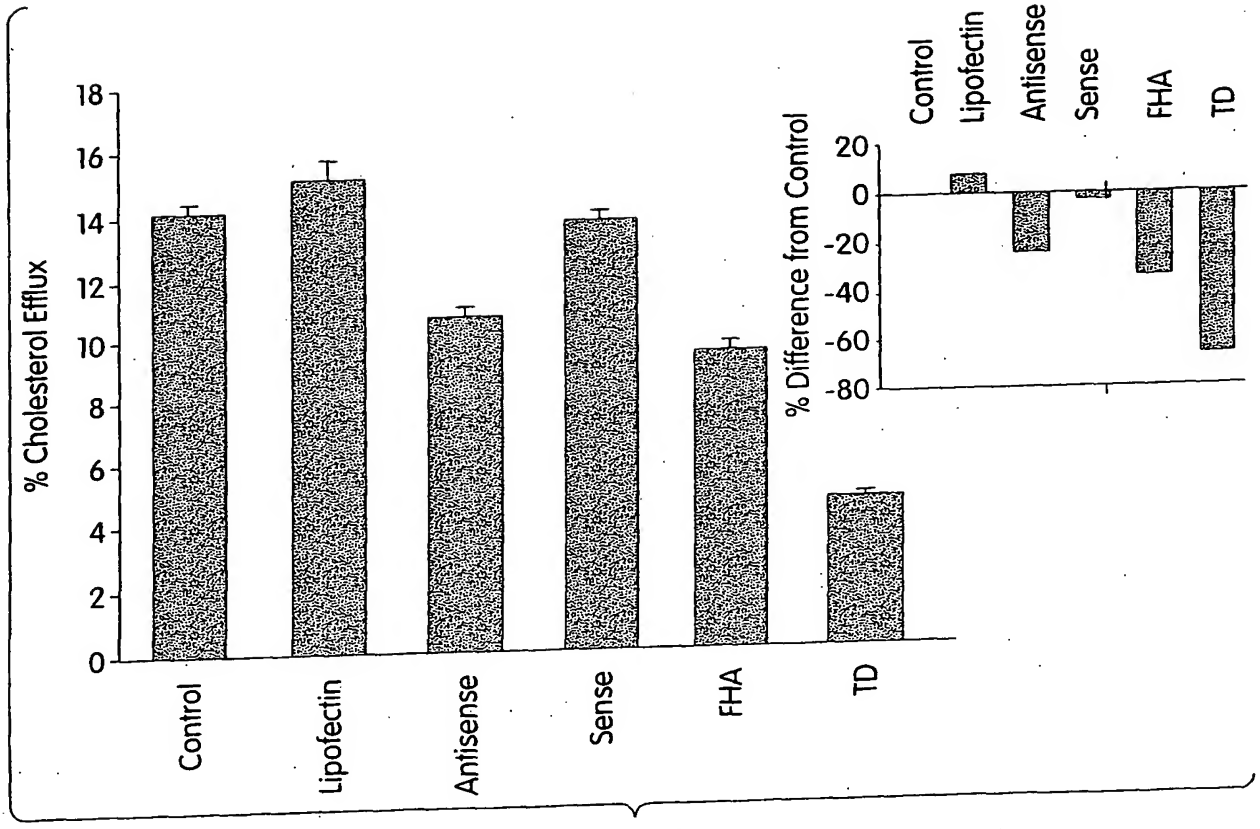
% Cholesterol Efflux
18
16
14
12
10
8
6
4
2
0
Control
Lipofectin
Antisense
Sense
FHA
TD
% Difference from Control
20
0
-20
-40
-60
-80
Control
Lipofectin
Antisense
Sense
FHA
TD

Fig. 7B

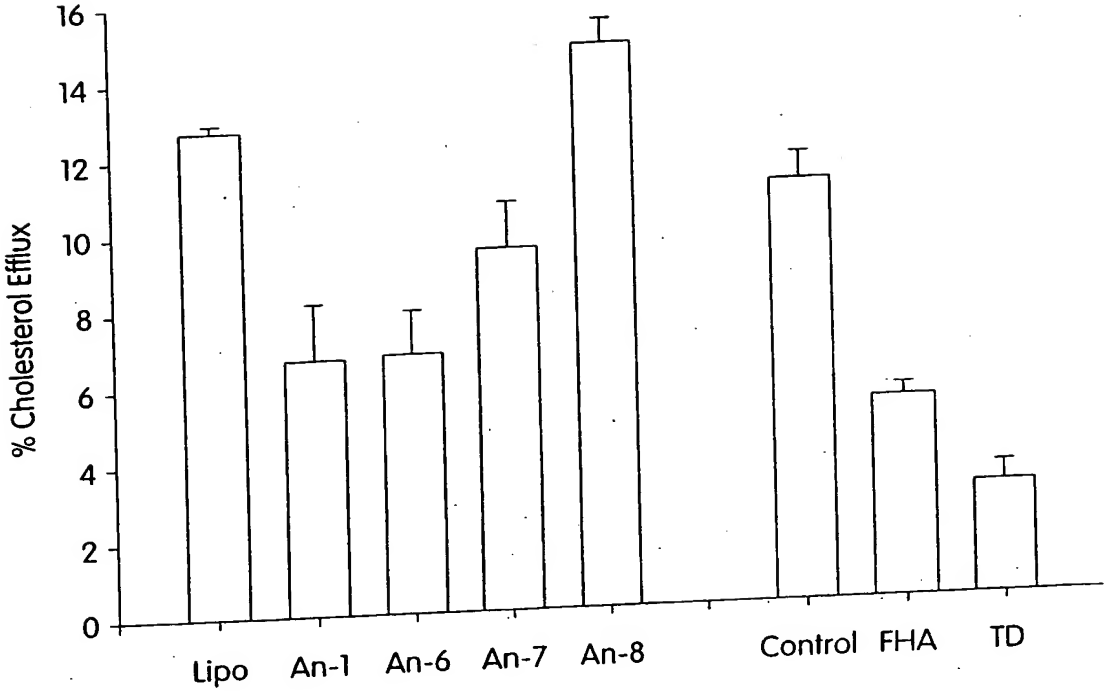
% Cholesterol Efflux
16
14
12
10
8
6
4
2
0
Lipo
An-1
An-6
An-7
An-8
Control
FHA
TD

Fig. 7C

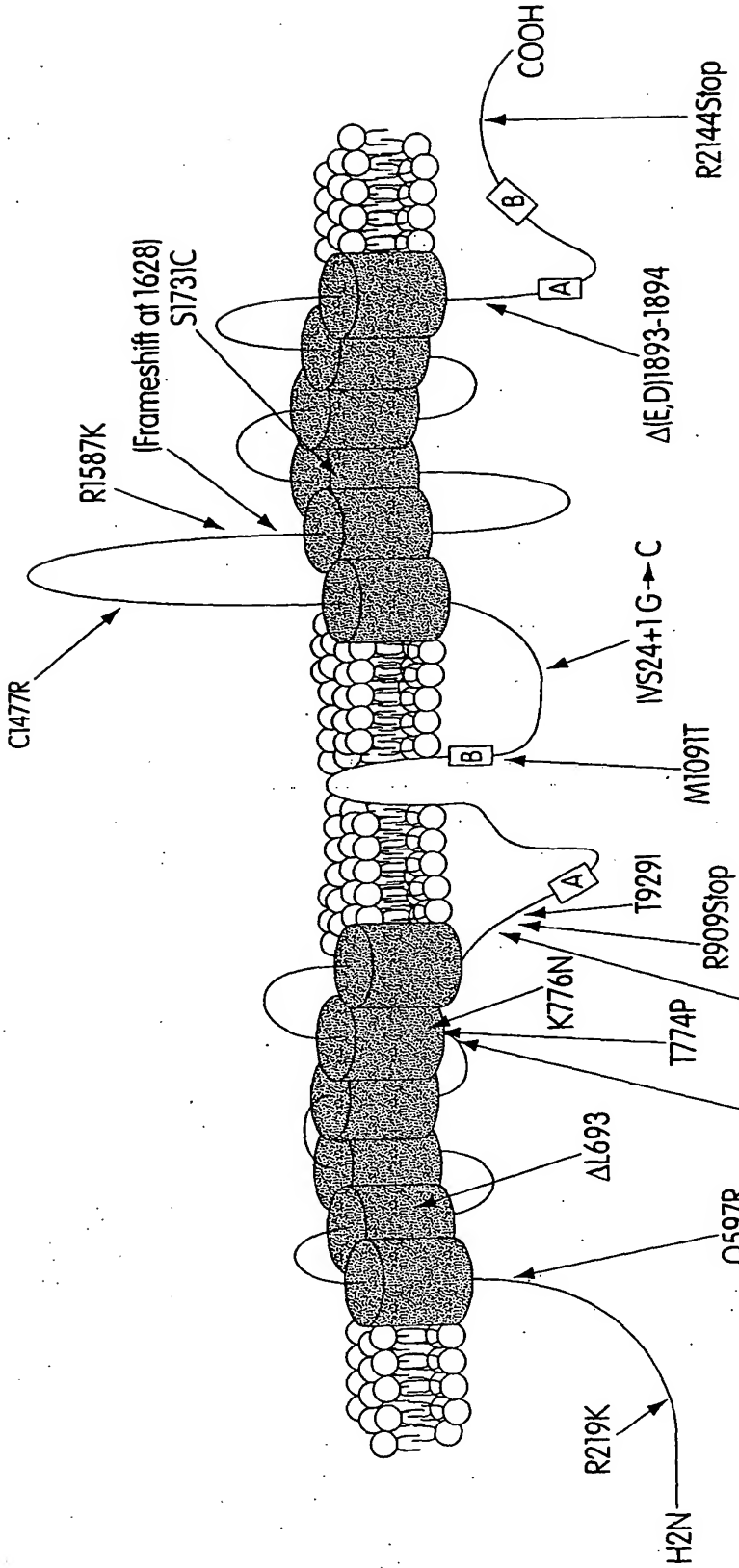
H2N
R219K
Q597R
ΔL693
V771M
T774P
K776N
1883M
R909Stop
T929I
M1091T
IVS24+1 G→C
C1477R
R1587K
(Frameshift at 1628)
S1731C
Δ(E,D)1893-1894
R2144Stop
COOH
A
B
A
B

Fig. 8

SEQ ID NO: 1

MACWPQLRLLLWKNLTFRRRQTCQLLLEVAWPLFIFLILISVRLSYPPYEQHECHFPNKAMPSAGTLPWVQ
GIICNANNPCFRYPTPGEAPGVVGNFNKSIVARLFSDARRLLLYSQKDTSMKDMRKVLRTLQQIKKSSSNL
KLQDFLVDNETFSGFLYHNLSLPKSTVDKMLRADVILHKVFLQGYQLHLTSLCNGSKSEEMIQLGDQEVSE
LCGLPREKLAAAERVLRSNMDILKPILRTLNSTSPFPSKELAEATKTLLHSLGTLAQELFSMRSWSDMRQE
VMFLTNVNSSSSSTQIYQAVSRIVCGHPEGGGLKIKSLNWYEDNNYKALFGGNGTEEDAETFYDNSTTPYC
NDLMKNLESSPLSRIIWKALKPLLVGKILYTPDTPATRQVMAEVNKTFQELAVFHDLEGMWEELSPKIWTF
MENSQEMDLVRMLLDSRDNDHFWEQQLDGLDWTAQDIVAFLAKHPEDVQSSNGSVYTWREAFNETNQAIRT
ISRFMECVNLNKLEPIATEVWLINKSMELLDERKFWAGIVFTGITPGSIELPHHVKYKIRMDIDNVERTNK
IKDGYWDPGPRADPFEDMRYVWGGFAYLQDVVEQAIIRVLTGTEKKTGVYMQQMPYPCYVDDIFLRVMSRS
MPLFMTLAWIYSVAVIIKGIVYEKEARLKETMRIMGLDNSILWFSWFISSLIPLLVSAGLLVVILKLGNLL
PYSDPSVVFVFLSVFAVVTILQCFLISTLFSRANLAAACGGIIYFTLYLPYVLCVAWQDYVGFTLKIFASL
LSPVAFGFGCEYFALFEEQGIGVQWDNLFESPVEEDGFNLTTSVSMMLFDTFLYGVMTWYIEAVFPGQYGI
PRPWYFPCTKSYWFGEESDEKSHPGSNQKRISEICMEEEPTHLKLGVSIQNLVKVYRDGMKVAVDGLALNF
YEGQITSFLGHNGAGKTTTMSILTGLFPPTSGTAYILGKDIRSEMSTIRQNLGVCPQHNVLFDMLTVEEHI
WFYARLKGLSEKHVKAEMEQMALDVGLPSSKLKSKTSQLSGGMQRKLSVALAFVGGSKVVILDEPTAGVDP
YSRRGIWELLLKYRQGRTIILSTHHMDEADVLGDRIAIISHGKLCCVGSSLFLKNQLGTGYYLTLVKKDVE
SSLSSCRNSSSTVSYLKKEDSVSQSSSDAGLGSDHESDTLTIDVSAISNLIRKHVSEARLVEDIGHELTYV
LPYEAAKEGAFVELFHEIDDRLSDLGISSYGISETTLEEIFLKVAEESGVDAETSDGTLPARRNRRAFGDK
QSCLRPFTEDDAADPNDSDIDPESRETDLLSGMDGKGSYQVKGWKLTQQQFVALLWKRLLIARRSRKGFFA
QIVLPAVFVCIALVFSLIVPPFGKYPSLELQPWMYNEQYTFVSNDAPEDTGTLELLNALTKDPGFGTRCME
GNPIPDTPCQAGEEEWTTAPVPQTIMDLFQNGNWTMQNPSPACQCSSDKIKKMLPVCPPGAGGLPPPQRKQ
NTADILQDLTGRNISDYLVKTYVQIIAKSLKNKIWVNEFRYGGFSLGVSNTQALPPSQEVNDAIKQMKKHL
KLAKDSSADRFLNSLGRFMTGLDTRNNVKVWFNNKGWHAISSFLNVINNAILRANLQKGENPSHYGITAFN
HPLNLTKQQLSEVALMTTSVDVLVSICVIFAMSFVPASFVVFLIQERVSKAKHLQFISGVKPVIYWLSNFV
WDMCNYVVPATLVIIIFICFQQKSYVSSTNLPVLALLLLLYGWSITPLMYPASFVFKIPSTAYVVLTSVNL
FIGINGSVATFVLELFTDNKLNNINDILKSVFLIFPHFCLGRGLIDMVKNQAMADALERFGENRFVSPLSW
DLVGRNLFAMAVEGVVFFLITVLIQYRFFIRPRPVNAKLSPLNDEDEDVRRERQRILDGGGQNDILEIKEL
TKIYRRKRKPAVDRICVGIPPGECFGLLGVNGAGKSSTFKMLTGDTTVTRGDAFLNKNSILSNIHEVHQNM
GYCPQFDAITELLTGREHVEFFALLRGVPEKEVGKVGEWAIRKLGLVKYGEKYAGNYSGGNKRKLSTAMAL
IGGPPVVFLDEPTTGMDPKARRFLWNCALSVVKEGRSVVLTSHSMEECEALCTRMAIMVNGRFRCLGSVQH
LKNRFGDGYTIVVRIAGSNPDLKPVQDFFGLAFPGSVLKEKHRNMLQYQLPSSLSSLARIFSILSQSKKRL
HIEDYSVSQTTLDQVFVNFAKDQSDDDHLKDLSLHKNQTVVDVAVLTSFLQDEKVKESYV*

Fig. 9A

SEQ ID NO: 2

GTCCCTGCTGTGAGCTCTGGCCGCTGCCTTCCAGGGCTCCCGAGCCACACGCTGGGGGTG
CTGGCTGAGGGAACATGGCTTGTTGGCCTCAGCTGAGGTTGCTGCTGTGGAAGAACCTCA
CTTTCAGAAGAAGACAAACATGTCAGCTGTTACTGGAAGTGGCCTGGCCTCTATTTATCT
TCCTGATCCTGATCTCTGTTCGGCTGAGCTACCCACCCTATGAACAACATGAATGCCATT
TTCCAAATAAAGCCATGCCCTCTGCAGGAACACTTCCTTGGGTTCAGGGGATTATCTGTA
ATGCCAACAACCCCTGTTTCCGTTACCCGACTCCTGGGGAGGCTCCCGGAGTTGTTGGAA
ACTTTAACAAATCCATTGTGGCTCGCCTGTTCTCAGATGCTCGGAGGCTTCTTTTATACA
GCCAGAAAGACACCAGCATGAAGGACATGCGCAAAGTTCTGAGAACATTACAGCAGATCA
AGAAATCCAGCTCAAACTTGAAGCTTCAAGATTTCCTGGTGGACAATGAAACCTTCTCTG
GGTTCCTGTATCACAACCTCTCTCTCCCAAAGTCTACTGTGGACAAGATGCTGAGGGCTG
ATGTCATTCTCCACAAGGTATTTTTGCAAGGCTACCAGTTACATTTGACAAGTCTGTGCA
ATGGATCAAAATCAGAAGAGATGATTCAACTTGGTGACCAAGAAGTTTCTGAGCTTTGTG
GCCTACCAAGGGAGAAACTGGCTGCAGCAGAGCGAGTACTTCGTTCCAACATGGACATCC
TGAAGCCAATCCTGAGAACACTAAACTCTACATCTCCCTTCCCGAGCAAGGAGCTGGCTG
AAGCCACAAAAACATTGCTGCATAGTCTTGGGACTCTGGCCCAGGAGCTGTTCAGCATGA
GAAGCTGGAGTGACATGCGACAGGAGGTGATGTTTCTGACCAATGTGAACAGCTCCAGCT
CCTCCACCCAAATCTACCAGGCTGTGTCTCGTATTGTCTGCGGGCATCCCGAGGGAGGGG
GGCTGAAGATCAAGTCTCTCAACTGGTATGAGGACAACAACTACAAAGCCCTCTTTGGAG
GCAATGGCACTGAGGAAGATGCTGAAACCTTCTATGACAACTCTACAACTCCTTACTGCA
ATGATTTGATGAAGAATTTGGAGTCTAGTCCTCTTTCCCGCATTATCTGGAAAGCTCTGA
AGCCGCTGCTCGTTGGGAAGATCCTGTATACACCTGACACTCCAGCCACAAGGCAGGTCA
TGGCTGAGGTGAACAAGACCTTCCAGGAACTGGCTGTGTTCCATGATCTGGAAGGCATGT
GGGAGGAACTCAGCCCCAAGATCTGGACCTTCATGGAGAACAGCCAAGAAATGGACCTTG
TCCGGATGCTGTTGGACAGCAGGGACAATGACCACTTTTGGGAACAGCAGTTGGATGGCT
TAGATTGGACAGCCCAAGACATCGTGGCGTTTTTGGCCAAGCACCCAGAGGATGTCCAGT
CCAGTAATGGTTCTGTGTACACCTGGAGAGAAGCTTTCAACGAGACTAACCAGGCAATCC
GGACCATATCTCGCTTCATGGAGTGTGTCAACCTGAACAAGCTAGAACCCATAGCAACAG
AAGTCTGGCTCATCAACAAGTCCATGGAGCTGCTGGATGAGAGGAAGTTCTGGGCTGGTA
TTGTGTTCACTGGAATTACTCCAGGCAGCATTGAGCTGCCCCATCATGTCAAGTACAAGA
TCCGAATGGACATTGACAATGTGGAGAGGACAAATAAAATCAAGGATGGGTACTGGGACC
CTGGTCCTCGAGCTGACCCCTTTGAGGACATGCGGTACGTCTGGGGGGGCTTCGCCTACT
TGCAGGATGTGGTGGAGCAGGCAATCATCAGGGTGCTGACGGGCACCGAGAAGAAAACTG

Fig. 9B

GTGTCTATATGCAACAGATGCCCTATCCCTGTTACGTTGATGACATCTTTCTGCGGGTGA
TGAGCCGGTCAATGCCCCTCTTCATGACGCTGGCCTGGATTTACTCAGTGGCTGTGATCA
TCAAGGGCATCGTGTATGAGAAGGAGGCACGGCTGAAAGAGACCATGCGGATCATGGGCC
TGGACAACAGCATCCTCTGGTTTAGCTGGTTCATTAGTAGCCTCATTCCTCTTCTTGTGA
GCGCTGGCCTGCTAGTGGTCATCCTGAAGTTAGGAAACCTGCTGCCCTACAGTGATCCCA
GCGTGGTGTTTGTCTTCCTGTCCGTGTTTGCTGTGGTGACAATCCTGCAGTGCTTCCTGA
TTAGCACACTCTTCTCCAGAGCCAACCTGGCAGCAGCCTGTGGGGGCATCATCTACTTCA
CGCTGTACCTGCCCTACGTCCTGTGTGTGGCATGGCAGGACTACGTGGGCTTCACACTCA
AGATCTTCGCTAGCCTGCTGTCTCCTGTGGCTTTTGGGTTTGGCTGTGAGTACTTTGCCC
TTTTTGAGGAGCAGGGCATTGGAGTGCAGTGGGACAACCTGTTTGAGAGTCCTGTGGAGG
AAGATGGCTTCAATCTCACCACTTCGGTCTCCATGATGCTGTTTGACACCTTCCTCTATG
GGGTGATGACCTGGTACATTGAGGCTGTCTTTCCAGGCCAGTACGGAATTCCCAGGCCCT
GGTATTTTCCTTGCACCAAGTCCTACTGGTTTGGCGAGGAAAGTGATGAGAAGAGCCACC
CTGGTTCCAACCAGAAGAGAATATCAGAAATCTGCATGGAGGAGGAACCCACCCACTTGA
AGCTGGGCGTGTCCATTCAGAACCTGGTAAAAGTCTACCGAGATGGGATGAAGGTGGCTG
TCGATGGCCTGGCACTGAATTTTTATGAGGGCCAGATCACCTCCTTCCTGGGCCACAATG
GAGCGGGGAAGACGACCACCATGTCAATCCTGACCGGGTTGTTCCCCCCGACCTCGGGCA
CCGCCTACATCCTGGGAAAAGACATTCGCTCTGAGATGAGCACCATCCGGCAGAACCTGG
GGGTCTGTCCCCAGCATAACGTGCTGTTTGACATGCTGACTGTCGAAGAACACATCTGGT
TCTATGCCCGCTTGAAAGGGCTCTCTGAGAAGCACGTGAAGGCGGAGATGGAGCAGATGG
CCCTGGATGTTGGTTTGCCATCAAGCAAGCTGAAAAGCAAAACAAGCCAGCTGTCAGGTG
GAATGCAGAGAAAGCTATCTGTGGCCTTGGCCTTTGTCGGGGGATCTAAGGTTGTCATTC
TGGATGAACCCACAGCTGGTGTGGACCCTTACTCCCGCAGGGGAATATGGGAGCTGCTGC
TGAAATACCGACAAGGCCGCACCATTATTCTCTCTACACACCACATGGATGAAGCGGACG
TCCTGGGGGACAGGATTGCCATCATCTCCCATGGGAAGCTGTGCTGTGTGGGCTCCTCCC
TGTTTCTGAAGAACCAGCTGGGAACAGGCTACTACCTGACCTTGGTCAAGAAAGATGTGG
AATCCTCCCTCAGTTCCTGCAGAAACAGTAGTAGCACTGTGTCATACCTGAAAAACGGAGG
ACAGTGTTTCTCAGAGCAGTTCTGATGCTGGCCTGGGCAGCGACCATGAGAGTGACACGC
TGACCATCGATGTCTCTGCTATCTCCAACCTCATCAGGAAGCATGTGTCTGAAGCCCGGC
TGGTGGAAGACATAGGGCATGAGCTGACCTATGTGCTGCCATATGAAGCTGCTAAGGAGG
GAGCCTTTGTGGAACTCTTTCATGAGATTGATGACCGGCTCTCAGACCTGGGCATTTCTA
GTTATGGCATCTCAGAGACGACCCTGGAAGAAATATTCCTCAAGGTGGCCGAAGAGAGTG
GGGTGGATGCTGAGACCTCAGATGGTACCTTGCCAGCAAGACGAAACAGGCGCGCCTTCG
GGGACAAGCAGAGCTGTCTTCGCCCGTTCACTGAAGATGATGCTGCTGATCCAAATGATT

Fig. 9C

CTGACATAGACCCAGAATCCAGAGAGACAGACTTGCTCAGTGGGATGGATGGCAAAGGGT
CCTACCAGGTGAAAGGCTGGAAACTTACACAGCAACAGTTTGTGGCCCTTTTGTGGAAGA
GACTGCTAATTGCCAGACGGAGTCGGAAAGGATTTTTTGCTCAGATTGTCTTGCCAGCTG
TGTTTGTCTGCATTGCCCTTGTGTTCAGCCTGATCGTGCCACCCTTTGGCAAGTACCCCA
GCCTGGAACTTCAGCCCTGGATGTACAACGAACAGTACACATTTGTCAGCAATGATGCTC
CTGAGGACACGGGAACCCTGGAACTCTTAAACGCCCTCACCAAAGACCCTGGCTTCGGGA
CCCGCTGTATGGAAGGAAACCCAATCCCAGACACGCCCTGCCAGGCAGGGGAGGAAGAGT
GGACCACTGCCCCAGTTCCCCAGACCATCATGGACCTCTTCCAGAATGGGAACTGGACAA
TGCAGAACCCTTCACCTGCATGCCAGTGTAGCAGCGACAAAATCAAGAAGATGCTGCCTG
TGTGTCCCCCAGGGGCAGGGGGGCTGCCTCCTCCACAAAGAAAACAAAACACTGCAGATA
TCCTTCAGGACCTGACAGGAAGAAACATTTCGGATTATCTGGTGAAGACGTATGTGCAGA
TCATAGCCAAAAGCTTAAAGAACAAGATCTGGGTGAATGAGTTTAGGTATGGCGGCTTTT
CCCTGGGTGTCAGTAATACTCAAGCACTTCCTCCGAGTCAAGAAGTTAATGATGCCATCA
AACAAATGAAGAAACACCTAAAGCTGGCCAAGGACAGTTCTGCAGATCGATTTCTCAACA
GCTTGGGAAGATTTATGACAGGACTGGACACCAGAAATAATGTCAAGGTGTGGTTCAATA
ACAAGGGCTGGCATGCAATCAGCTCTTTCCTGAATGTCATCAACAATGCCATTCTCCGGG
CCAACCTGCAAAAGGGAGAGAACCCTAGCCATTATGGAATTACTGCTTTCAATCATCCCC
TGAATCTCACCAAGCAGCAGCTCTCAGAGGTGGCTCTGATGACCACATCAGTGGATGTCC
TTGTGTCCATCTGTGTCATCTTTGCAATGTCCTTCGTCCCAGCCAGCTTTGTCGTATTCC
TGATCCAGGAGCGGGTCAGCAAAGCAAAACACCTGCAGTTCATCAGTGGAGTGAAGCCTG
TCATCTACTGGCTCTCTAATTTTGTCTGGGATATGTGCAATTACGTTGTCCCTGCCACAC
TGGTCATTATCATCTTCATCTGCTTCCAGCAGAAGTCCTATGTGTCCTCCACCAATCTGC
CTGTGCTAGCCCTTCTACTTTTGCTGTATGGGTGGTCAATCACACCTCTCATGTACCCAG
CCTCCTTTGTGTTCAAGATCCCCAGCACAGCCTATGTGGTGCTCACCAGCGTGAACCTCT
TCATTGGCATTAATGGCAGCGTGGCCACCTTTGTGCTGGAGCTGTTCACCGACAATAAGC
TGAATAATATCAATGATATCCTGAAGTCCGTGTTCTTGATCTTCCCACATTTTTGCCTGG
GACGAGGGCTCATCGACATGGTGAAAAACCAGGCAATGGCTGATGCCCTGGAAAGGTTTG
GGGAGAATCGCTTTGTGTCACCATTATCTTGGGACTTGGTGGGACGAAACCTCTTCGCCA
TGGCCGTGGAAGGGGTGGTGTTCTTCCTCATTACTGTTCTGATCCAGTACAGATTCTTCA
TCAGGCCCAGACCTGTAAATGCAAAGCTATCTCCTCTGAATGATGAAGATGAAGATGTGA
GGCGGGAAAGACAGAGAATTCTTGATGGTGGAGGCCAGAATGACATCTTAGAAATCAAGG
AGTTGACGAAGATATATAGAAGGAAGCGGAAGCCTGCTGTTGACAGGATTTCGCGTGGGCA
TTCCTCCTGGTGAGTGCTTTGGGCTCCTGGGAGTTAATGGGGCTGGAAAATCATCAACTT
TCAAGATGTTAACAGGAGATACCACTGTTACCAGAGGAGATGCTTTCCTTAACAAAAATA

Fig. 9D

GTATCTTATCAAACATCCATGAAGTACATCAGAACATGGGCTACTGCCCTCAGTTTGATG
CCATCACAGAGCTGTTGACTGGGAGAGAACACGTGGAGTTCTTTGCCCTTTTGAGAGGAG
TCCCAGAGAAAGAAGTTGGCAAGGTTGGTGAGTGGGCGATTCGGAAACTGGGCCTCGTGA
AGTATGGAGAAAAATATGCTGGTAACTATAGTGGAGGCAACAAACGCAAGCTCTCTACAG
CCATGGCTTTGATCGGCGGGCCTCCTGTGGTGTTTCTGGATGAACCCACCACAGGCATGG
ATCCCAAAGCCCGGCGGTTCTTGTGGAATTGTGCCCTAAGTGTTGTCAAGGAGGGGAGAT
CAGTAGTGCTTACATCTCATAGTATGGAAGAATGTGAAGCTCTTTGCACTAGGATGGCAA
TCATGGTCAATGGAAGGTTCAGGTGCCTTGGCAGTGTCCAGCATCTAAAAAATAGGTTTG
GAGATGGTTATACAATAGTTGTACGAATAGCAGGGTCCAACCCGGACCTGAAGCCTGTCC
AGGATTTCTTTGGACTTGCATTTCCTGGAAGTGTTCTAAAAGAGAAACACCGGAACATGC
TACAATACCAGCTTCCATCTTCATTATCTTCTCTGGCCAGGATATTCAGCATCCTCTCCC
AGAGCAAAAAGCGACTCCACATAGAAGACTACTCTGTTTCTCAGACAACACTTGACCAAG
TATTTGTGAACTTTGCCAAGGACCAAAGTGATGATGACCACTTAAAAGACCTCTCATTAC
ACAAAAACCAGACAGTAGTGGACGTTGCAGTTCTCACATCTTTTCTACAGGATGAGAAAG
TGAAAGAAAGCTATGTATGAAGAATCCTGTTCATACGGGGTGGCTGAAAGTAAAGAGGAA
CTAGACTTTCCTTTGCACCATGTGAAGTGTTGTGGAGAAAAGAGCCAGAAGTTGATGTGG
GAAGAAGTAAACTGGATACTGTACTGATACTATTCAATGCAATGCAATTCAATGCAATGA
AAACAAAATTCCATTACAGGGGCAGTGCCTTTGTAGCCTATGTCTTGTATGGCTCTCAAG
TGAAAGACTTGAATTTAGTTTTTTACCTATACCTATGTGAAACTCTATTATGGAACCCAA
TGGACATATGGGTTTGAACTCACACTTTTTTTTTTTTTTTTGTTCCTGTGTATTCTCATT
GGGGTTGCAACAATAATTCATCAAGTAATCATGGCCAGCGATTATTGATCAAAATCAAAA
GGTAATGCACATCCTCATTCACTAAGCCATGCCATGCCCAGGAGACTGGTTTCCCGGTGA
CACATCCATTGCTGGCAATGAGTGTGCCAGAGTTATTAGTGCCAAGTTTTTCAGAAAGTT
TGAAGCACCATGGTGTGTCATGCTCACTTTTGTGAAAGCTGCTCTGCTCAGAGTCTATCA
ACATTGAATATCAGTTGACAGAATGGTGCCATGCGTGGCTAACATCCTGCTTTGATTCCC
TCTGATAAGCTGTTCTGGTGGCAGTAACATGCAACAAAAATGTGGGTGTCTCCAGGCACG
GGAAACTTGGTTCCATTGTTATATTGTCCTATGCTTCGAGCCATGGGTCTACAGGGTCAT
CCTTATGAGACTCTTAAATATACTTAGATCCTGGTAAGAGGCAAAGAATCAACAGCCAAA
CTGCTGGGGCTGCAACTGCTGAAGCCAGGGCATGGGATTAAAGAGATTGTGCGTTCAAAC
CTAGGGAAGCCTGTGCCCATTTGTCCTGACTGTCTGCTAACATGGTACACTGCATCTCAA
GATGTTTATCTGACACAAGTGTATTATTTCTGGCTTTTTGAATTAATCTAGAAAATGAAA

Fig. 9E

	Exon (bp)	Forward Primer	SEQ ID No.	Reverse Primer	SEQ ID No.			intron(kb)	intron (kb)
exon 1	140	GGCTGGATTAGCAGTCCTCA	70	ATCCCCAACTCAAAACCACA	119	intron	1	>6.413	>6.4
exon 2	94	GGATTTCCCAGATCCCAGTG	71	AAGTCCAATTTAGCCCACGTT	120	intron	2	>4.241	>4.2
exon 3	142	GACAGACTTGGCATGAAGCA	72	CCAGCCATTCAAAATTCTCC	121	intron	3	>1.248(1.6)	1.6
exon 4	119	GCACTTGGCAGTCACTTCTG	73	GGGTGCAGGTCAATTTCCAAT	122	intron	4	>1.512	>1.5
exon 5	122	CGTTTCTCCACTGTCCCATT	74	CCCCTTCACCACCATTACAA	123	intron	5	>1.796(3)	3
exon 6	177	ACTTCAAGGACCCAGCTTCC	75	TGTCCAAGGAAAAGCCTCAC	124	intron	6	>2.726 (10)	10
exon 7	93	TCGGTTTCTTGTTTGTTAAACTCA	76	AGGACCTCTTGCCAGACTCA	125	intron	7	4.975	5
exon 8	241	TCCCAAGGCTTTGAGATGAC	77	AGGAGATGACACAGGCCAAG	126	intron	8	>2.311(.5)	2.5
exon 9	140	GGCTCCAAAGCCCTTGTAA	78	CGCACACCTCTGAAGCTACC	127	intron	9	0.332	0.3
exon 10	117	GCTGCTGTGATGGGGTATCT	79	ACCTCACTCACACCTGGGAA	128	intron	10	4.208	4.2
exon 11	198	TTTGTAAATTTTGTAGTGCTCCTCA	80	GCCTCCTGCCTGAACCTTAT	129	intron	11	0.747	0.7
exon 12	206	TAGTCAGCCCTTGCCTCCTA	81	CAAAATCATGACACCAAGTTGAG	130	intron	12	0.523	0.5
exon 13	177	AAAGGGGCTTGGTAAGGGTA	82	CATGCACATGCACACACATA	131	intron	13	1.787	1.8
exon 14	223	GATGTGGTGCTCCCTCTAGC	83	CCTTAGCCCGTGTTGAGCTA	132	intron	14	1.747	1.7
exon 15	222	CAAGTGAGTGCTTGGGATTG	84	TGCTTTTATTCAGGGACTCCA	133	intron	15	1.059	1.1
exon 16	205	GCAAATTCAAATTTCTCCAGG	85	CCCATGCACTGCAGAGATTC	134	intron	16	1.105	1.1
exon 17	114	TCAAGGAGGAAATGGACCTG	86	AAGGCAGGAGACATCGCTT	135	intron	17	1.789	1.8
exon 18	172	CTGAAAGTTCAAGCGCAGTG	87	GGGATCAGCATGGTTTCCTA	136	intron	18	0.99	1
exon 19	132	TGCAGACTGAATGGAGCATC	88	GCTTAAGTCCCACTCCTCCC	137	intron	19	1.307	1.3
exon 20	143	GCCAGGGGACACTGTATTCT	89	ATTTTCCTCCGCATGTGTGT	138	intron	20	0.204	0.2
exon 21	138	AGGTCCTCTGCCTTCACTCA	90	TCACAGAAGCCTAGCCATGA	139	intron	21	0.706	0.7
exon 22	221	CCAGTGCTTACCCCTGCTAA	91	AACAGAGCAGGGAGATGGTG	140	intron	22	>0.866(1.7)	1.7
exon 23	73	CACACAACAGAGCTTCTTGGA	92	TCTGCACCTCTCCTCCTCTG	141	intron	23	0.986	1
exon 24	203	ACCTGGAACAGGTGTGGTGT	93	ACTGGGGCCAACATTAATCA	142	intron	24	1.668	1.7
exon 25	49	GGGCTAACATGCCACTCAGTA	94	CTTCCCCATCTGCAACAAAC	143	intron	25	0.196	0.2

Figure 10A

exon 26	114	GTTTGTTGCAGATGGGGAAG	95	GCTAAAGGCCATCCAAAGAA	144	intron 26	1.396	1.4
exon 27	149	CACCAGAAGAAGGAGCATGG	96	TCAAGTGCATCTGGGCATAA	145	intron 27	1.649	1.6
exon 28	125	CTGGACTCGTAGGGATTTGC	97	TCTGAAGTCCATTCCCTTGG	146	intron 28	>0.728(1.4)	1.4
exon 29	99	GCCTGTCACAGAGAAATGCTT	98	CAATGTGGCATGCAGTTGAT	147	intron 29	>2.589(3)	3
exon 30	190	TTACGGAATGATCCTGTGCTC	99	GAAGCTACCAGCCCATCCT	148	intron 30	1.521	1.5
exon 31	95	AGTCAGGTTTCCGGTCACAC	100	CATTTCCCCCACTGTTTCAG	149	intron 31	>0.944(\)	>0.9
exon 32	33	CCGTTCCTTATATCCTCAGGTG	101	CCAAGGCTTTCTTCAATCCA	150	intron 32	>1.062(/6.5)	>1.0
exon 33	106	CCTTGTACACACTCGCACTGA	102	GATCCGTTTAACCTGCCAAC	151	intron 33	1.475	1.5
exon 34	75	TGTTGTCCACAGGTTCCAGA	103	ATGCCCCTGCCAACTTTAC	152	intron 34	0.522	0.5
exon 35	170	TGAGGTTTATGGGCATGGTT	104	CTCTGCAGCTGTTCCCCTAC	153	intron 35	1.228	1.2
exon 36	178	ATGTTTTTCCTTGGCTGTGC	105	TATCAATCCATGGCCCTGAC	154	intron 36	>1.898(2)	2
exon 37	116	ATCTGCCCTTTCTTGTCTGA	106	AGAGTCCCTGCCCTCCTTCT	155	intron 37	0.112	0.1
exon 38	145	AGGGAGCTGCACAGTGGATA	107	AAGGCAGTCAGCAGTGTCAA	156	intron 38	1.545	1.5
exon 39	124	TCACTCCCATATTTCAGAACTTGA	108	GGGGAACATCCTGTGCTTAG	157	intron 39	1.087	1.1
exon 40	130	TGTTTATTGGAAGATCGGTGAA	109	CCATTGGTGAGTGTTTCCCT	158	intron 40	0.265	0.3
exon 41	121	CGTTAGAGACTGAATCTTTGTCCTG	110	AGTCAGCAAACTGCTGGGTT	159	intron 41	>0.622(0.9)	0.9
exon 42	63	AGTCCTGCCTTCCACAGTTG	111	ATTGCTCCATCCTGGCATAA	160	intron 42	0.909	0.9
exon 43	107	GGTAGTTACGTGTTAGGGGCA	112	TCATGGATGATTTTATGTGCTTC	161	intron 43	2.355	2.4
exon 44	142	CAGGAACATTAGGCCAGATTG	113	GCGTGTGGAAAAGCCATAAG	162	intron 44	0.372	0.4
exon 45	135	CATGTATGTGTAGGACAGCATGA	114	GCCAATCATACAACAGCCCT	163	intron 45	>1.059(1.3)	1.3
exon 46	104	CTGTTTCAAAGATGCTTCTGC	115	TGATCGCATATTCTACTTGGAAA	164	intron 46	0.483	0.5
exon 47	93	CCTAGGAAGCTGGAATGCTG	116	TCCCTTTATTTTAGAGGCACCA	165	intron 47	0.659	0.7
xon 48	244	GGGTTCCCAGGGTTCAGTAT	117	GATCAGGAATTCAAGCACCAA	166	intron 48	0.941	0.9
exon 49	295	CTTGACCTAATTTCAACATCTGG	118	TGGGTTCCATAATAGAGTTTCACA	167		>1.075	

Figure 10B

ERRORS IN PUBLIC SEQUENCE (differences between samples and Genbank entry AJ012376.1):					
Exon/Inrton	Nucleotides	Amino Acid Change		Sequence difference/context	SEQ ID NO:
2	T150C A152G	no change	Public sequence: Correct sequence:	TGTCAGCTGTTACTGGAAGTGG TGTCAGCTGCTGCTGGAAGTGG	168 169
7	C839T	no change	Public sequence: Correct sequence:	AGGAGCTGGCCGAAGCCACAA AGGAGCTGGCTGAAGCCACAA	170 171
33	C4738T	T1495I	Public sequence: Correct sequence:	AATGATGCCACCAAACAAATG AATGATGCCATCAAACAAATG	172 173
35	C5017T	P1588L	Public sequence: Correct sequence:	GAGGTGGCTCCGATGACCACA GAGGTGGCTCTGATGACCACA	174 175
43	G5995A	R1914K	Public sequence: Correct sequence:	TTCCTTAACAGAAATAGTATC TTCCTTAACAAAAATAGTATC	176 177
48	C6577T	P2108L	Public sequence: Correct sequence:	GGAAGTGTTCCAAAAGAGAAA GGAAGTGTTCTAAAAGAGAAA	178 179
49	G6899A	not applicable	Public sequence: Correct sequence:	AGTAAAGAGGGACTAGACTTT AGTAAAGAGGAACTAGACTTT	180 181
Mutations:					SEQ ID NO:
13	A1864G	Q597R	More common: Less common:	GCCTACTTGCAGGATGTGGTG GCCTACTTGCGGGATGTGGTG	182 183
14	delta CTT 2151-3	deltaL093	More common: Less common:	CCTCATTCCTCTTCTTGTGAGCG CCTCATTCCT/CTTGTGAGCG	184 185
15	G2385A	V771M	More common: Less common:	GCAGGACTACGTGGGCTTCAC GCAGGACTACATGGGCTTCAC	186 187
18	C2799T	R909Stop	More common: Less common:	AAAAGTCTACCGAGATGGGAT AAAAGTCTACTGAGATGGGAT	188 189
18	C2860T	T929I	More common: Less common:	GGCCAGATCACCTCCTTCCTG GGCCAGATCATCTCCTTCCTG	190 191
22	T3346C	M1091T	More common: Less common:	ACACACCACATGGATGAAGCG ACACACCACACGGATGAAGCG	192 193

Figure 11A

Intron 24	(+1) G to C splice donor site	Altered transcript lenght	More common: Less common:	CCTGGAAGAAGTAAGTTAAGT CCTGGAAGAACTAAGTTAAGT	194 195
30	T4503C	C1477R	More common: Less common:	GCTGCCTGTGTGTCCCCCAGG GCTGCCTGTGCGTCCCCCAGG	196 197
35	GG 4958-57 to C	frameshift at aa 1628	More common: Less common:	TAGCCATTATGGAATTACTGCT TAGCCATTATCAATTACTGCT	198 199
41	delta AAGATG 5752-7	delta(E.D)1893-1894	More common: Less common:	GATGAAGATGAAGATGTGAGGCGGGA GATGAAGATG/TGAGGCGGGA	200 201
48	C6504T	R2144Stop	More common: Less common:	AATAGTTGTACGAATAGCAGG AATAGTTGTATGAATAGCAGG	202 203
Promoter Variants:					
Location	Position Relative to Xenon cDNA	Position Realitive to SEQ ID NO: 14 Containing Exon 1			SEQ ID NO:
1	G57C	8216	More common: Less common:	ACACGCTGGGGGTGCTGGCTG ACACGCTGGGGGTGCTGGCTG	204 205
5	(-)4 ins. G	8158	More common: Less common:	GACCAGCCACGGCGTCCCTG GACCAGCCACGGGCGTCCCTG	206 207
5	A (-)380 G	7780	More common: Less common:	CATTTTCTTAGAAAAGAGAGGT CATTTTCTTAGAGAAGAGAGGT	208 209
5	A (-)479 C	7681	More common: Less common:	GAAAATTAGTATGTAAGGAAG GAAAATTAGTCTGTAAGGAAG	210 211
5	A (-)738 G	7422	More common: Less common:	CCTCCGCCTGCCAGGTTCAGCGATT CCTCCGCCTGCCGGGTTCAGCGATT	212 213
5	A (-)1045 G	7115	More common: Less common:	TATGTGCTGACCATGGGAGCTTGTT TATGTGCTGACCGTGGGAGCTTGTT	214 215
5	A (-)1113 G	7047	More common: Less common:	GTGACACCCAACGGAGTAGGG GTGACACCGAGCGGAGTAGGG	216 217
5	(-)1181 ins. CCCT	6979	More common: Less common:	AGTATCCCT/TGTTCACGAGAA AGTATCCCTCCCTTGTTCACGAGAA	218 219

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Figure 11B

Polymorphisms:					
Exon/Inrton	Nucleotides	Amino Acid Change		Sequence difference/context	SEQ ID NO:
5	G548A	no change	More common: Less common:	CTGGGTTCCTGTATCACAACC CTGGGTTCCTATATCACAACC	220 221
6	G730A	R219K	More common: Less common:	GGCCTACCAAGGGAGAAACTG GGCCTACCAAAGGAGAAACTG	222 223
Intron 7	G(+)2383 T	Not applicable	Allele 1: Allele 2:	TTTAAAGGGGGTGATTAGGA TTTAAAGGGGTTGATTAGGA	224 225
Intron 7	G(+)3035 T	Not applicable	Allele 1: Allele 2:	GAAGAATTTGTTTTTTGATT GAAGAATTTTTTTTTTTGATT	226 227
8	C1010T	no change	More common: Less common:	GCGGGCATCCCGAGGGAGGGG GCGGGCATCCTGAGGGAGGGG	228 229
8	G1022A	no change	More common: Less common:	AGGGAGGGGGGCTGAAGATCA AGGGAGGGGGACTGAAGATCA	230 231
Intron 9	(-)42 ins. G	Not applicable	More common: Less common:	AGGAGCCAAACGCTCATTGT AGGAGCCAAAGCGCTCATTGT	232 233
Intron 13	T(+)24 A	Not applicable	More common: Less common:	AAGCCACTGTTTTTAACCAGT AAGCCACTGTATTTAACCAGT	234 235
15	A2394C	T774P	More common: Less common:	CGTGGGCTTCACACTCAAGAT CGTGGGCTTCCCACTCAAGAT	236 237
15	G2402C	K776N	More common: Less common:	TCACACTCAAGATCTTCGCTG TCACACTCAACATCTTCGCTG	238 239
Intron 14	C(+)16 T	Not applicable	Allele 1: Allele 2:	GCAGCCTCACCCGCTCTTCCC GCAGCCTCACTCGCTCTTCCC	240 241
17	A2723G	I883M	Allele 1: Allele 2:	AGAAGAGAATATCAGAAATCT AGAAGAGAATGTCAGAAATCT	242 243
Intron 17	C(+)2000 G	Not applicable	Allele 1: Allele 2:	GCGCAGTGCCCTGTGTCCTTA GCGCAGTGCGCTGTGTCCTTA	244 245

Figure 11C

21	T3233G	no change	More common:	GATCTAAGGTTGTCATTCTGG	246
			Less common:	GATCTAAGGTGGTCATTCTGG	247
Intron 21	G(+)118 T	Not applicable	Allele 1:	CTCTTCTGTTAGGACAGAAGAGA	248
			Allele 2:	CTCTTCTGTTATCACAGAAGAGA	249
Intron 21	A(+)563 G	Not applicable	Allele 1:	CATTCTAGGGATCATAGCCAT	250
			Allele 2:	CATTCTAGGGGTCATAGCCAT	251
Intron 24	G(+)321 T	Not applicable	Allele 1:	AAGTACAGTGGGAGGAACAGCG	252
			Allele 2:	AAGTACAGTGTGAGGAACAGCG	253
Intron 29	A(-)624 G	Not applicable	Allele 1:	AATTCCTAAAAAATAGAAATGCA	254
			Allele 2:	ATTCCTAAAAAAGTAGAAATGCA	255
Intron 31	T(+)30 C	Not applicable	More common:	GGCCCCTGCCTTATTATTACT	256
			Less common:	GGCCCCTGCCGTATTATTACT	257
Intron 33	A(+)732 G	Not applicable	Allele 1:	TGAGAGAATTACTTGAACCCGG	258
			Allele 2:	TGAGAGAATTGCTTGAACCCGG	259
Intron 33	C(+)898 T	Not applicable	Allele 1:	TTTGCTGAAACAATCACTGCA	260
			Allele 2:	TTTGCTGAAATAATCACTGAC	261
Intron 34	C(+) 234 T	Not applicable	Allele 1:	AACCTCAGTTCCCTCATCTGTG	262
			Allele 2:	AACCTCAGTTTCCTCATCTGTG	263
34	G4834A	R158TK	More common:	CTGGACACCAGAAATAATGTC	264
			Less common:	CTGGACACCAAAAATAATGTC	265
37	C 5266G	S1731C	More common:	TCCTATGTGTCCTCCACCAAT	266
			Less common:	TCCTATGTGTGCTCCACCAAT	267
Intron 43	T(+)18 C	Not applicable	More common:	AAGAAGTGGCTTGTATTTTGC	268
			Less common:	AAGAAGTGGCCTGTATTTTGC	269
Intron 43	C(+)1665 G	Not applicable	Allele 1:	AACTGATTTGATTGGTATAGCTG	270
			Allele 2:	AACTGATTTGGTTGGTATAGCTG	271
48	C6521T	no change	More common:	CAGGGTCCAACCCGGACCTGA	272
			Less common:	CAGGGTCCAATCCGGACCTGA	273
Intron 10	(+)14 ins. T	Not applicable	More common:	GCGTCAGGGATGGGGACAG	284
			Less common:	GCGTCAGGGATTGGGGACAG	285
Exon 16	G2547A	V8251	More common:	CCACTTCGGTCTCCATG	286
			Less common:	CCACTTCGATCTCCATG	287
Polymorphism in an ABC1 BAC contig; This polymorphism is within approximatey 200kb of the ABC1 gene					SEQ ID NO:
	A or G	Not applicable	Allele 1:	TTGGGAGGCTAAGGCAGGAGAA	274
			Allele 2:	TTGGGAGGCTGAGGCAGGAGAA	275

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Figure 11D

Genomic contig containing ABC1 exon 1:
Underline = putitive promotor lement

acctcttatagaatgatagaattcctctggaatgattggataacttcatttcatccttgacttttaccttggaggattt
cttacccctttggcttctcaaatttgactattaaaatgttgcctttaaaaataggaacacagtttcaggggggagtac
cagcccatgacccttctgcaaggcccctaactcaaggtagtttccctggaactgtggtttatggaatgtttcaggagt
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aagttaaactgaggcagtcagattagcttcaggctcaattaagctgatgggtcagcctgggagaaattgcaggatgact
ctcaatatcccctcccaccccacagcagccacgatctgtctgtctttaatcatgggtgcagtgaacctgttctttcca
ggtgtcttggccttcagtaaccttgttaggcttgtccctgaacgtggctaccgatccaaagacacatgatcagagaggc
aattagagaacagacctttccaaagcaagcatgttctgttgggcttagaagtttcatgtcctaatattataggaccct
gtgcatctctctggagatgaggcacatgagtcatatctgtgattcttgcttttgtgtcaacatctcatgaataggcaat
cagagctttggcaccaatgtattttcagttcatatctgatgtagttaaatccacctcctgctttgtagtttactggcaa
gctgtttttgatataagacatctagaacactgtaaatatataacatttttatttgtctattatacctcaattacgaaaa
agacatctagaagcaacctcatcaagagagatactgaggccgggcatggtagctcacacttgcaatcccattactttgg
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aatacgacagtcaaatggagcatgactttgtggaagcctctcctcttccacccagaggggccaatttctctgtcccagt
gagatgttgacacttgtatgatccctgcttggagacttcccctcttctggaacctgccctggctcaggcatgagggctga
ctgtcacccttcgataggagcccagcactaaagctcatgtgttggcagtgttcttgcgggaaggaaaaagaccagccag
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taatgaatatttatctactattcctcttccaaggcgatcacacaataatcaggctttacactatccagttcttaggtct
tccaagttatgacttgtgaggtatgttaattatgataatagaaggcagtttatttggttcagatttattgatgtgtaat
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Figure 12A-(1)

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atggtagaagctgctgaacacagagctccctctttggggataatttgcccaaatcatttaatcaggcttgagaaatgag
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gacatccccaagtgcttacgacaagccaggacccttttgcatactaaggaaaacagggatgaaggaaacagaaatggtc
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Figure 12A-(2)

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Figure 12A-(3)

SEQ ID NO: 15
Genomic contig containing ABC1 exon 2:

ctttttttttttttttttttttttttttgaggtgaagtctcactctgttgcccaggctggagtgcaatggagcgatc
ttggctcaccccaacctctgtctcctgggttcaaacagttctcctgcctcagcctcccgagtagctgggattacaggctc
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Figure 12B - (1)

actggagtagcacaggaggagggcttctagctcaggctgagattttagtaaaggaaattatgccacgatgaatcctgaag
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GCTGAGCTACCCACCCTATGAACAACATGAATgtaagtaactgtggatgttgcctgagactcaccaatggcagggaaaat
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attaaaagcacatttaagataatgccttaacacctagtcttttccatatgcatgatgtcttaatcacacattgcaaatca
tggaacacagaatttt

Figure 12B - (2)

SEQ ID NO 16
Genomic contig containing ABC1 exon 3:

atcttacaatcacagtctttctcttagggctgggctcagtgggtggattgacactgcagaaatggccagatctaaaggat
caacatttacgtagctgggaaatgtagctgggacttcagtttcactgccctagtgatttttcctaccactaagcagctca
gtccataccctacgagacccacaagcttatgagatactgttcttccaggaaagcagtggggccagggccacctttaat
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Figure 12C - (1)

cagggtttcgccatgttggccagggtggtcttgaactcctgacctcaggtgatccgcccaccttggcctcccaaagtgct
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Figure 12C - (2)

SEQ ID NO 17
Genomic contig containing ABC1 exon 4:

tcatgactgccattggtataaagatgaatataatccagaccagattcatgattattcatacatttttagtgtattaactt
ttaattctgcttttaaaataaattaaaacattctaatatgcccttaagagtatcccagcccaggccactgagcctactgt
ggttcatggataagtttgcccctgggggcatgtgtgtgcatgcatgtgtgtgcacatgcatgatgagccgggccttgaag
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AGTTCTGAGAACATTACAGCAGATCAAGAAATCCAGCTCAAgtaagtaaaaaccttctctgcatccgtttataattggaa
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ctgccatagcatcagagcagccttccaggcagtggcctggcaaggggacagaggctggtgggagcagctggctgagtgca
gccagtaatggcatgt

Figure 12D

SEQ ID NO 18
Genomic contig containing ABC1 exon 5:

agctctccaggtgattctgatgcatacttaagtttgagaaccattgcttgttttgcattaaacaggagattagtctctgc
agcttgtgggaataaagctttaaatctctccaattttagctctgtgaaaaggcagtggggagacaggaatgaacggacta
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Figure 12E

SEQ ID NO 19
Genomic contig containing ABC1 exon 6:

agtaaaatggagaattccaaattctgaaattgttagaacatagttctgtgtcttagttaaatatcgacacttacagataa
atagcataaatgctttctccccatatttcagcccagtcctacttaaagacaacataaattgcaaaatagtgaggatgttg
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GCAATGGATCAAAATCAGAAGAGATGATTCAACTTGGTGACCAAGAAGTTTCTGAGCTTTGTGGCCTACCAAGGGAGAAA
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Figure 12F

SEQ ID NO 20
Genomic contig containing ABC1 exon 8:

ccgtttggcaaatgctcagtaaaagaaaagggttagaaggggagaaaggcattttatcccaagccttcaggaatcaggat
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Figure 12G - (1)

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Figure 12G - (2)

cagaatccgcatttccaacaatgtctccagtaatgctgatgctgctcgtccctggaccacagattgggtagccaggttct
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Figure 12G - (3)

SEQ ID NO 21
Genomic contig containing ABC1 exon 9 through 22:

actgtgttagcaaggatggtctcgatctcctgacctcgtgatccgcctgtatcggcctcccaaagtgctgggattacagg
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Figure 12H - (1)

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Figure 12H - (2)

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Figure 12H - (3)

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Figure 12H - (4)

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Figure 12H - (5)

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AGCTATCTGTGGCCTTGGCCTTTGTCGGGGGATCTAAGGTTGTCATTCTGGATGAACCCACAGCTGGTGTGGACCCTTAC
TCCCGCAGGGGAATATGGGAGCTGCTGCTGAAATACCGACAAGgtgcctgatgtgtatttattctgagtaaatggactga
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kcacagaagagataaagggcattgagactgagattcctgagaggagatgctgtgtctttattcatctttttgtccccaac
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gctctctggttagagatctgatgtggtacagtatgaggagcacaggcaggcttggagccaactctggcttggccctgaga
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tctcttccagtgcttacccctgctaatatctctctagtccctgtcactgtgacaaatcagaactgagaggcctcacctgt
cccacatccttgtgtttgtgcctggcagGCCGCACCATTATTCTCTCTACACACCACATGGATGAAGCGGACGTCCTGGG
GGACAGGATTGCCATCATCTCCCATGGGAAGCTGTGCTGTGTGGGCTCCTCCCTGTTTCTGAAGAACCAGCTGGGAACAG
GCTACTACCTGACCTTGGTCAAGAAAGATGTGGAATCCTCCCTCAGTTCCTGCAGAAACAGTAGTAGCACTGTGTCATAC
CTGAAAAAGgtgagctgcagtcttggagctgggctggtgttgggtctgggcagccaggacttgctggctgtgaatgattt
ctccatctccacccctttgccatgttgaaaccaccatctccctgctctgttgccccctttgaaatcatatcatacttaag
gcatggaaagctaaggggccctctgctcccattgtgctagttctgttgaatcccgtttcctttcctatgaggcacana
gagtgatggagaaggtccttagaggacattattatgtcaaagaaaagagacttgtcaagaggtaagagccttggctacaa
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ggcactgatttcatgccagagacactctgggcattgaaagaaagtaatgataatagttaattttatatagcgttgttacc
atttcaacctttttttttttaacctctatcatctcaattaaag

Figure 12H - (6)

SEQ ID NO: 22
Genomic contig containing ABC1 exon 23 to 28:

gtgaacacacattaaagcatgagaagcatgaactagacatgtagccaggtaaaggccttgctgagatggttggcaaaggc
ctcattgcagcattcattggcaggccacagttcttttggcagctctgcttcctgacctttcaccctcaggaagcgaggct
gttcacacggcacacacatgccagacagggtcctctgaagccacggctgccagtgcatgtgtcccagggaaagctttttc
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cccgttctcacctccttctttgccatcagGAGGACAGTGTTTCTCAGAGCAGTTCTGATGCTGGCCTGGGCAGCGACCAT
GAGAGTGACACGCTGACCATCGgtaaggactctggggtttcttattcaggtggtgcctgagcttcccccagctgggcaga
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ggggtgggagcagcttggtggcgggttggcctaatgcttgctggggtgcctggggctcggtttgggagctagcagggcag
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catcctcccatcctcccattcacccatccacccatccagctgtccacccattctacactgagtacctataatgtgcctgg
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acctaaacacctggaacaggtgtggtgtatgagcggcaggcctctgatgagagggtgggggatggccagccctcactccg
aagcccctctgagttgattgagccatctttgcattctggtcctgcagATGTCTCTGCTATCTCCAACCTCATCAGGAAGC
ATGTGTCTGAAGCCCGGCTGGTGGAAGACATAGGGCATGAGCTGACCTATGTGCTGCCATATGAAGCTGCTAAGGAGGGA
GCCTTTGTGGAACTCTTTCATGAGATTGATGACCGGCTCTCAGACCTGGGCATTTCTAGTTATGGCATCTCAGAGACGAC
CCTGGAAGAAgtaagttaagtggctgactgtcggaatatatagcaaggccaaatgtcctaaggccagaccagtagcctgc
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atttaagatggtagtgggagatagcaggaaagaagtgttttcctctgtaccacagtacatgcctgagatttgtgtgttga
aaccagtggtacctaacacatttacatcccaaccttaaactcctatgcacttatttacccttaatgagcctctttactt
aagtacagtgkgaggaacagcggcatcaggatcacttgggaacttgttagaaattcagcaacttgggcccagctcagacc
tactgaatcagaatcaggagcaattctctggtgtgactgtgtcacagccaggtatcaactggattctcatacataggaaa
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taaacatactaaggtttggcttcagggcctgtgaatcagaatttctggaggtgaggccttgaagtctgtatttctattgc
atactttggacacagtggtctatagactagagtttggaaatgattgcgctcattcagattctcttctgatgtttgaattg
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gatttagagccacagtcctgaaagaagccacttgactcattccctgtaggttcagaataaatttcttctgcgcagtgtct
gtcatagctttttttaaatttttttttattttgatgagactggagttttgctcttattgcccaagctggagtgcagtgg
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caagcatgtgctaccacgcccagctaattttgtatttttagtagagatgggttttatccatgttggtcaggctggtctcg
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aatataataatcgtggcagtggtgactactctcgtaatgttggtgcttataatgttctcatctctctcatttccagATA
TTCCTCAAGGTGGCCGAAGAGAGTGGGGTGGATGCTGAGACCTCAGgtaactgccttgagggagaatggcacacttaaga
tagtgccttctgctggctttctcagtgcacgagtattgttcctttccctttgaattgttctattgcattctcatttgtag
agtgtaggtttgttgcagatggggaaggtttgttttgttgtaaataaaataaagtatgggattctttccttgtgccttca

Figure 12 I - (1)

gATGGTACCTTGCCAGCAAGACGAAACAGGCGGGCCTTCGGGGACAAGCAGAGCTGTCTTCGCCCGTTCACTGAAGATGA
TGCTGCTGATCCAAATGATTCTGACATAGACCCAGgtctgttagggcaagatcaaacagtgtcctactgtttgaatgtga
aattctctctcatgctctcacctgttttctttggatggcctttagccaaggtgatagatccctacagagtccaaagagaa
gtgaggaaatggtaaaagccacttgttctttgcagcatcgtgcatgtgatcaaacctgaaagagcctatccatatcactt
cctttaaagacataaagatggtgcctcaatcctctgaacccatgtatttattatcttttctgcggggtcctagtttcttg
tatacattaggtgtttaattgttgaacaaatattcattcgagtagatgagtgattttgaaagagtcagaaaggggaattt
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gagaggctgaggtgggtagatcacctgaggtcaggagtttgagaccagtctgaccaacaaggtgaaaccccgtctctact
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gaacccaggaggcagaggttgcagtcagccacggttgcgccattgcactccagactgggcaacaagagtgaaaactccat
ctcaaaaaagaaaaaaaaagaattagatattttggatgagtgtgtctttgtgtgtttaactgagatggagaggagagcta
agacatcaaacaaatattgttaagatgtaaaagcacatcagttaggtatcattagtttaggacaaggatttctagaaaat
ttttaggaacagaaaactttccagttctctcacccctgctcaaagagtgtatggctcttacattatatataactgcctga
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ctaattgccattgtgtattctcaaatatgtcaagctacgtacatggcctgtttcatagagtagtctataagaaattgatg
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tttctgtacttatcagacaaggactgaaaggcagagactgctactgttagacattttgagtcaagctttccttggacat
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agaagaaggagcatggcaatgccgaaaacacctaagataataggtgaccttcagtgttggcttcttgcagAATCCAGAGA
GACAGACTTGCTCAGTGGGATGGATGGCAAAGGGTCCTACCAGGTGAAAGGCTGGAAACTTACACAGCAACAGTTTGTGG
CCCTTTTGTGGAAGAGACTGCTAATTGCCAGACGGAGTCGGAAAGGATTTTTTGCTCAGgtgagacgtgctgttttcgcc
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cggagccagatttatctgtatttcaatcccagttctattccttgccagttgtgtatccgctggcaagttacttctctatg
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caggctccattgcttcccagctgccaagaatgccttggtgcagcacagtcataggccctgcattcctcattgccgtgctg
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cctgccccctccactgcctgggtaaagATTGTCTTGCCAGCTGTGTTTGTCTGCATTGCCCTTGTGTTCAGCCTGATCGT
GCCACCCTTTGGCAAGTACCCCAGCCTGGAACTTCAGCCCTGGATGTACAACGAACAGTACACATTTGTCAGgtatgttt
gtcttctacatcccaggaggggtaagattcgagcagaccaaagatgtttacgagggccaagggaatggacttcagaatt
acacggtggaat

Figure 12 I - (2)

SEQ ID NO: 23
Genomic contig containing ABC1 exon 29:

gggaagcatttaaaaaaaaaaagtatatatatatatatatatatatatatgtaatgtgaattggcctctttttctctaa
gcccacattttcttcttacatagttcaggtttactttatttttccttccggctgctgaccctgtattgcccgtagttg
tggaacatagcatgtgtttgtgacctgtgcctgttatttttgtgctttctagttgtgcatgcaaagagtacaaagttttc
ttgccctttcttggaaaatcctgcttgtctgtgccaaagggataattgtgaaagcacttttgaaatacttaatgagttga
ttttcttcaaattaaaaaaaatatataaatgtatatgtgtatgtacatgtgtgtacacatacacacctttatacatacag
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aacatttttatctctcgattactggttttaatgaattactaagctggctgcctctcatgtacccacagCAATGATGCTCC
TGAGGACACGGGAACCCTGGAACTCTTAAACGCCCTCACCAAAGACCCTGGCTTCGGGACCCGCTGTATGGAAGGAAACC
CAATCCCgtgagtgccactttagccataagcagggcttcttgtgcttgttgcctggtttgatttctaatatgctgcattt
atcaactgcatgccacattgtgaccgccagcatttgccctttgaattattattatgttttatttacaaaaagcgaaggta
gtaaccgaactaaattatctaggaacaaacgtttggagagtcttctaacaccgyscaaagcacgtcattacagacatttg
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cccagcccctgtacttaagtgcttcaataggctctcattatatatgatttttaggttttgcttatcagcttcttcgcttt
tataatctgaaaagatggcatatgaatttttataaaaagggacactttcttcttctcaaattgtatatttttattgtact
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taggggaagggaggtcaccagatcactgtgagtgaagatggtggagaggtgaggatcttatgaggccgtgctcaaggctg
gtagaggtgggttagtgtttccaggtttaggcagaatctcagctgaggtcatgaaacaacagtgatctctgaaaaattat
ggcaaggtgggaaggtgctggagaattggagaggggcaaacttgactttcaagtttcaatgggaagataggtgactctg
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ctaatattggttcccaaataaccagatgacaaggatagagaaggacacagacacggcctatctggatttcatggtgcctt
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gccagctgaaccatatgaactcatggattgatgctagcttaggaaggctctgtaggagccagaactgggctgagagccag
cccatagagacaaaagaggcccggccctgacatcagagggttcaaacatgatgtctgagccccacctacagtctgccgga
ggtggttggaaggaagagcctttatccttacaattcttactgaaattcaaattttaggttttgcaaaaaaatggtggac
ctgaaggaaatttgacaggagcatgtctcagctgtatttaaatttgtctcagccaatccccttttgaatgttcagagtgt
aagcttcaggagggcagcgcgtcttagtgtgacttttctggtcagttcaggtgctttaaggagacaattagagatcaatc
tggaaaacttcatttgaatttttaatacataagaaaacaataagaaatagttaaaaatatatatttatataatatatata
tgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtatatatatatatatttttatttatttatttttttttgagatggagtctcg
ctctgttgcccaggctggagtgcagtggctcaatcttggctcactgccacctctgcctcccaggttcaagtgattctcct
acctcagcctcctgagtagctgggattacaagcatgtgccaccacactggctaa

Figure 12J

SEQ ID NO: 24
Genomic contig containing ABC1 exon 30 and 31:

tcttgccagtctctactcatttttcagcacatcgagcataagatccagactctttcccaggcctctctcatctggctcct
ctcctcctcctttatcattactcttcttcgtagcttatcctactccagccatgctgtcttcctattattcctaaaaarta
gaaatgcatttcttcctagggcctttgtacctgcacttgccatcgcttttgctcagaatgttctttttgccaagcttttg
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tttattttgctttatttttgggccatctaaggttatcttattagtgtatttgttgttcgtctcctccatgggcatacacct
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aaatgagatcatgcttgtcttttccaaatggtgagggaatagatgcatgtttgtggttgttacggaatgatcctgtgctc
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CAGGGGAGGAAGAGTGGACCACTGCCCCAGTTCCCCAGACCATCATGGACCTCTTCCAGAATGGGAACTGGACAATGCAG
AACCCTTCACCTGCATGCCAGTGTAGCAGCGACAAAATCAAGAAGATGCTGCCTGTGTGTCCCCCAGGGGCAGGGGGGCT
GCCTCCTCCACAAgtgagtcactttcaggggtgattgggcagaaggggtgcaggatgggctggtagcttccgcttggaa
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agggtttcgccgtgttggctaggctggtctggaattcctgacctcaggtgatccacccgcctcggcctcccaaagtgctg
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caggtgcggggtggctcacacctgtaatctcagtattttgggatgccgaggtggactgatcacctaaggtcaggagttcg
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AGACGTATGTGCAGATCATAGCCAAAAGgtgacttttactaaacttggcccctgccttattattactaattagaggaat
taaagacctacaaataacagactgaaacagtgggggaaatgccagattatggcctgattctgtctattggaagtttagga
tattatcccaaactagaaaagatgacgagagggactgtgaacattcagttgtcagcttcaaggctgaggcagcctggtct
agaatgaaaatagaaatggattcaacgtcaaattttgccac

Figure 12K

SEQ ID NO: 25
Genomic contig containing ABC1 exon 32:_

gcatgctggagtgatagtgaccatgagtttctaagaaagaagcataatttctccatatgtcatccacaattgaaatatta
ttgttaattgaaaaagcttctaggccaggcacggtggctcatgcctgtaatcccagcactttaggagccaaggcgggtgg
atcacttgaggtcaggagtttgagaccagcctggccaacatggggaaaccctgtctctactaaaaatacaaaataagctg
ggcgtggtggtgcgtgcctgtaatcccagctacttgggaggctgaggcaggagaactgcttgaatctgggaggcggaggt
tgcagtgagctgagttcatgccattgcattccagcctgggcaacaagagcgaaaccatctcccaaaagaaaaaaaaaaga
aagaaaaagcttctagtttggttacatcttggtctataaggtggtttgtaaattggtttaacccaaggcctggttctcat
ataagtaatagggtatttatgatggagagaaggctggaagaggcctgaacacaggcttcttttctctagcacaaccctac
aaggccagctgattctagggttatttctgtccgttccttatatcctcaggtggatatttactccttttgcatcattagga
ataggctcagtgctttctttgaactgatttttgtttctttgtctctgcagCTTAAAGAACAAGATCTGGGTGAATGAGT
TTAGgtaagttgctgtctttctggcacgtttagctcaggggggaggatggtgttgtaggtgtgcttggattgaagaaagcc
ttggggattgtttgtcactcacacacttgtgggtgccatctcactgtgagga

Figure 12L

SEQ ID NO: 26
Genomic contig containing ABC1 exon 33 to 36:

gctttatagagtttctgcctagagcatcatggctcagtgcccagcagcccctccagaggcctctgaatatttgatatact
gatttccttgaggagaatcagaaatctcctgcaggtgtctagggatttcaagtaagtagtgttgtgaggggaatacctac
ttgtactttccccccaaaccagattcccgaggcttcttaaggactcaaggacaatttctaggcatttagcacgggactaa
aaaggtcttagaggaaataagaagcgccaaaaccatctctttgcactgtatttcaacccatttgtccttctgggttttga
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TAATACTCAAGCACTTCCTCCGAGTCAAGAAGTTAATGATGCCATCAAACAAATGAAGAAACACCTAAAGCTGGCCAAGg
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aacagaccctaggattgtggacagcagtgatagtcctagagtccacaagtctgcttttgagtgatgggcccatgtatctg
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AGGACTGGACACCAGAAATAATGTCAAGgtaaaccgctgtctttgttctagtagctttttgatgaacaataatccttatg
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tgggtatgcacctgcctgcaggagcccggtttgggggcattaccttgtacctggtatgttttctttcagGTGTGGTTCAA
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gtaggggaacagctgcagagaccgttatattatgataaatctggattgatccaggctctgggcagaagtgataagtttac
gaattggctggttgggcttcttgaactgcagaagagaaaatgacactgatatgtaaaaatcgtaacatttagtgaattca

Figure 12M – (1)

tataaagtgagttcaaaaattgttaattaaattataatttaattataagtgtttaatcagtttgatttgtttaaaaacca
ctgttttaaatttggtggaatatgtttttattagcttgtatctttaattcctaaattaagctgtgtgtgtgtgtgtgtgt
gtgtgtgtgtgtgtgtgtgtgtgtgtgtgaagtttaaagccaggatgagctagtttaaagtatgcagcctttggagtc
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gcccagcactcagcacagagttgatatttcacacacattagataccttcctgtatgtggagcatggcagttcctgtttc
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tctgtgtgtgtgtacgtgtgtgtttgtctgtgtgtccatgtcctcactgattgagccctaactgcatcaaagacccctca
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ctaattcttctcccaccgaacaagtccctggatatttaaaaataactctcatactctcatttaacctgagtattacccag
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Figure 12M – (2)

SEQ ID NO: 27
Genomic contig containing ABC1 exon 37 to 41:

aaattactctgactgggaatccatcgttcagtaagtttactgagtgtgacaccttggcttgactgttggaaagacagaaa
gggcatgtagtttataaaatcagccaaggggaaaatgcttgtcaaaatgtattgtcgggtattttgattaatagtttatg
tggcttcattaattcagagttactctccaatatgtttatctgccctttcttgtctgataatggtgaaaacttgtgtgatg
cattgtatatttgatttaggggtgaactggatgtctttgttttcacttttagTGCAATTACGTTGTCCCTGCCACACTGG
TCATTATCATCTTCATCTGCTTCCAGCAGAAGTCCTATGTGTCCTCCACCAATCTGCCTGTGCTAGCCCTTCTACTTTTG
CTGTATGGgtaagtcacctctgagtgagggagctgcacagtggataaggcatttggtgcccagtgtcagaaggagggcag
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TCAAGATCCCCAGCACAGCCTATGTGGTGCTCACCAGCGTGAACCTCTTCATTGGCATTAATGGCAGCGTGGCCACCTTT
GTGCTGGAGCTGTTCACCGACAATgtgagtcatgcagagagaacactcctgctgggatgagcatctctgggagccagagg
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CGTGTTCTTGATCTTCCCACATTTTTGCCTGGGACGAGGGCTCATCGACATGGTGAAAAACCAGGCAATGGCTGATGCCC
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aagggccatggtttttgatccctccattctgaccggatctctgcattgtgtctactagGCGAGAATCGCTTTGTGTCACC
ATTATCTTGGGACTTGGTGGGACGAAACCTCTTCGCCATGGCCGTGGAAGCGGTGGTGTTCTTCCTCATTACTGTTCTGA

Figure 12N – (1)

TCCAGTACAGATTCTTCATCAGGCCCAGgtgagcttttcttagaacccgtggagcacctggttgagggtcacagaggag
gcgcacagggaaacactcaccaatgggggttgcattgaactgaactcaaaatatgtgataaaactgattttcctgatgtg
ggcatcccgcagccccctccctgcccatcctggagactgtggcaagtaggttttataatactacgttagagactgaatct
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AATGATGAAGATGAAGATGTGAGGCGGGAAAGACAGAGAATTCTTGATGGTGGAGGCCAGAATGACATCTTAGAAATCAA
GGAGTTGACGAAGgtgagagagtacaggttacaatagctcatcttcagtttttttcagctttatgtgctgtaacccagca
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aacaccatggagacttcttaaagaattcttactgagaattcttttgtgatatgaattcccattctcgaatactttggttt
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gatt

Figure 12N – (2)

SEQ ID NO: 28
Genomic contig containing ABC1 exon 42 to 45:

ttttaaaatacctgcaatacatatatatgttgaatagatgaaaaattatgtagatgataatgaatgatacggttctaaaa
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gctcttggcacatcaaatatgcctatggcttagggttattgacaagtcttatgttgcagtgtatgtggtttatagtcctg
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GGAAGCGGAAGCCTGCTGTTGACAGGATTTGCGTGGGCATTCCTCCTGGTGAGgtaaagacactttgtctatattgcgtt
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aatacaatttaagaaaaagcatggggctgggcgcagtgattcactcctgtaatcccagcactttgggaggccgaggtggg
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cacatctaaaacatgcttttgtgatccatttgggatggtgatgacattcaaatagtttttaaaaatagattttctcctt
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ggtttgaggtagttacgtgttaggggcatttatattcatgtttttagagtttgcttatacaacttaatctttccttttca
gTGCTTTGGGCTCCTGGGAGTTAATGGGGCTGGAAAATCATCAACTTTCAAGATGTTAACAGGAGATACCACTGTTACCA
GAGGAGATGCTTTCCTTAACAAAAATAGgtgagaaaagaagtggcttgtattttgctgcaaagactttgtttttaattta
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gttggccagactggtctcgagctcctgacctcaggtgatccacctgcctcagcctcccaaagttctgggattacaggcat
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Figure 120 – (1)

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aaaggctgtgctttcaagtagcagcagatgtattggtatctttgtaatggagaagcatactttacaggaacattaggcca
gattgtctaaccagagtatctctacctgcttaaaatctaagtagttttcttgtcctttgcagTATCTTATCAAACATCCA
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ggaatttcaccccaatcttatactgacttcaatagaggtttcagacaaaaagttgttttgtat

Figure 12O – (2)

SEQ ID NO: 29
Genomic contig containing ABC1 exon 46 to 49:

ngccnngttnaaaangaaaatttnnnnnaaattnaannttanngngnnnttccccagaaaaaacnaaaangatttccn
cccngggggngccccnantcnaaaaggccccnctttnttgnggngagggaaagnttttttggaatttttaatttttgg
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Figure 12P – (1)

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Figure 12P – (2)

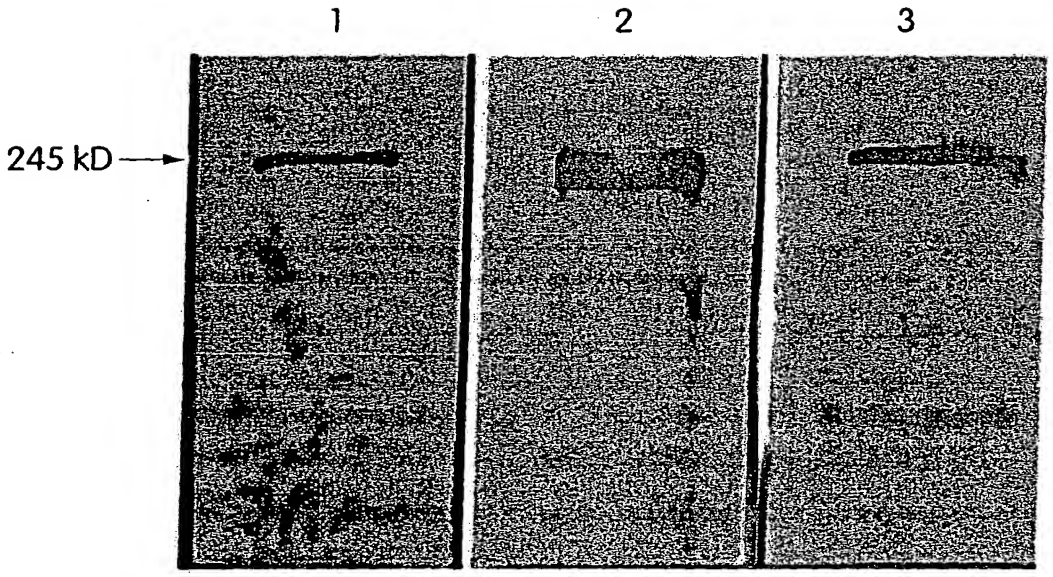
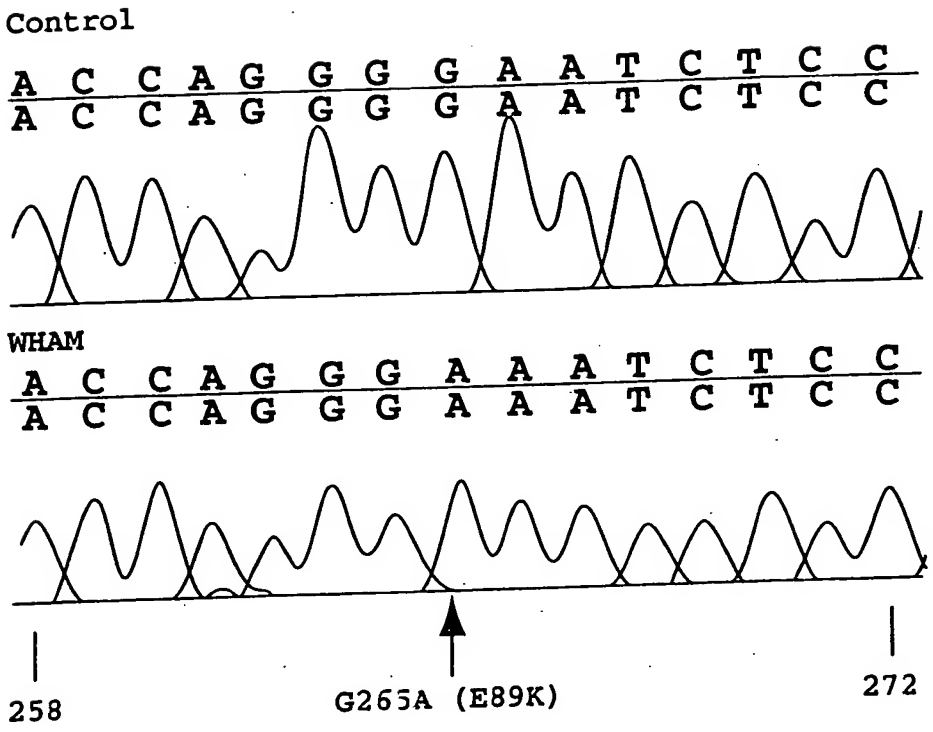


Fig. 13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A C C A G G G G A A T C T C C
WHAM
A C C A G G G A A A T C T C C
A C C A G G G A A A T C T C C
258
G265A (E89K)
272

Fig. 14

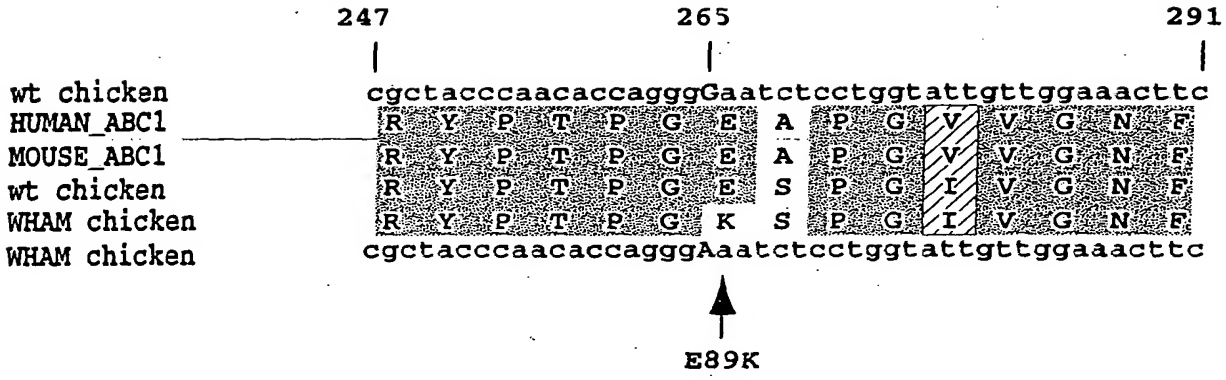
247
265
291
wt chicken cgctacccaacaccagggGaatctcctggtattgttggaaacttc
HUMAN_ABC1 R Y P T P G E A P G V V G N F
MOUSE_ABC1 R Y P T P G E A P G V V G N F
wt chicken R Y P T P G E S P G I V G N F
WHAM chicken R Y P T P G K S P G I V G N F
WHAM chicken cgctacccaacaccagggAaatctcctggtattgttggaaacttc
E89K

Fig. 15

No.	Name	Location in SEQ ID No. 14	Sequence	Sequence Length	Strand
1	PPRE	58-69	AGGTAAAAGTCA	12	Complement
2	PPRE	1997-2009	AGAGTAGAGGGCA	13	Lead
3	PPRE	2150-2161	ATGTCAAGTTCA	12	Lead
4	PPRE	2156-2169	AGTTCAAAAGGGCA	14	Lead
5	PPRE	4126-1139	AGGCCAGCAGGGCC	14	Complement
6	PPRE	5075-5087	AGGGCAGAAGTGA	13	Lead
7	PPRE	6604-6615	ATGCCAAGGTCA	12	Complement
8	PPRE	6731-6743	GGGGCAAGGGTA	13	Complement
9	PPRE	7220-7233	AGGTAATGAGGACA	14	Complement
10	PPRE	7554-7568	GGATCACGAGGTCA	15	Complement
1	SRE	159-166	CAGCCCAT	8	Lead
2	SRE	1133-1140	CAGCTCAC	8	Complement
3	SRE	1145-1152	CACACCAC	8	Lead
4	SRE	1809-1816	CAGCCCTC	8	Complement
5	SRE	1894-1901	CAGCCCAT	8	Lead
6	SRE	2563-2570	CAACCCAC	8	Lead
7	SRE	3303-3310	CAGCTCAC	8	Lead
8	SRE	3470-3477	CCGCCCAC	8	Lead
9	SRE	4784-4791	CTCCCCAC	8	Complement
10	SRE	4802-4809	CAGCCTAC	8	Complement
11	SRE	4970-4977	CACCTCAC	8	Complement
12	SRE	6487-6494	CAGCCTAC	8	Complement
13	SRE	6565-6572	CACCCAAC	8	Complement
14	SRE	6727-6734	CACCCTCA	8	Lead
15	SRE	7041-7048	CACCCAAC	8	Lead
16	SRE	8059-8066	CAGCCCTC	8	Complement
1	ROR(retinoic acid receptor related)	166-172	AGGGTCA	7	Complement
2	ROR(retinoic acid receptor related)	166-173	AAGGGTCA	8	Complement
3	ROR(r tinoic acid receptor related)	263-370	ATGGGTCA	8	Lead
4	ROR(retinoic acid receptor related)	264-370	TGGGTCA	7	Lead
5	ROR(r tinoic acid receptor related)	2218-2225	TAGGGTCA	8	Lead
6	ROR(retinoic acid receptor related)	2219-2225	AGGGTCA	7	Lead
7	ROR(retinoic acid receptor related)	3643-2649	TGGGTCA	7	Lead
8	ROR(retinoic acid receptor related)	6604-6610	AAGGTCA	7	Complement
1	SREBP-1 or "E box"	473-479	ACACCTG	7	Complement
2	SREBP-1 or "E box"	536-541	ACACATG	7	Lead
3	SREBP-1 or "E box"	537-543	TCATGTG	7	Complement
4	SREBP-1 or "E box"	655-661	TCATGTG	7	Complement
5	SREBP-1 or "E box"	925-931	ACACTTG	7	Lead
6	SREBP-1 or "E box"	967-973	TCACTTG	7	Lead
7	SREBP-1 or "E box"	968-974	TCAAGTG	7	Complement
8	SREBP-1 or "E box"	1053-1069	ACAGGTG	7	Complement
9	SREBP-1 or "E box"	1104-1110	TCACTTG	7	Lead
10	SREBP-1 or "E box"	1105-1111	TCAAGTG	7	Complement
11	SREBP-1 or "E box"	1561-1567	TCACTTG	7	Lead

Figure 16A

12	SREBP-1 or "E box"	1670-1676	TCAAATG	7	Lead
13	SREBP-1 or "E box"	1748-1754	ACACTTG	7	Lead
14	SREBP-1 or "E box"	1749-1755	ACAAGTG	7	Complement
15	SREBP-1 or "E box"	1852-1858	TCATGTG	7	Lead
16	SREBP-1 or "E box"	1853-1859	ACACATG	7	Complement
17	SREBP-1 or "E box"	1899-1905	ACAAATG	7	Complement
18	SREBP-1 or "E box"	2199-2205	ACACGTG	7	Lead
19	SREBP-1 or "E box"	2393-2399	ACAGCTG	7	Complement
20	SREBP-1 or "E box"	2669-27005	ACACCTG	7	Lead
21	SREBP-1 or "E box"	2677-2683	TCACATG	7	Complement
22	SREBP-1 or "E box"	2740-2746	ACAACTG	7	Complement
23	SREBP-1 or "E box"	2969-5975	ACAAATG	7	Lead
24	SREBP-1 or "E box"	2979-2985	ACACATG	7	Lead
25	SREBP-1 or "E box"	2981-2987	ACATGTG	7	Lead
26	SREBP-1 or "E box"	2980-2986	ACATGTG	7	Complement
27	SREBP-1 or "E box"	2982-2988	ACACATG	7	Complement
28	SREBP-1 or "E box"	3461-3467	TCAGGTG	7	Lead
29	SREBP-1 or "E box"	3462-2468	TCACCTG	7	Complement
30	SREBP-1 or "E box"	3547-3553	TCAACTG	7	Complement
31	SREBP-1 or "E box"	3752-3758	ACACATG	7	Lead
32	SREBP-1 or "E box"	4226-4232	TCACCTG	7	Lead
33	SREBP-1 or "E box"	4582-4588	ACACGTG	7	Complement
34	SREBP-1 or "E box"	4588-4594	TCAGTTG	7	Lead
35	SREBP-1 or "E box"	4861-4867	TCAGGTG	7	Lead
36	SREBP-1 or "E box"	4951-4957	ACAAATG	7	Lead
37	SREBP-1 or "E box"	5096-5102	TCAAATG	7	Complement
38	SREBP-1 or "E box"	5912-5918	ACAGTTG	7	Lead
39	SREBP-1 or "E box"	5913-5919	TCAACTG	7	Complement
40	SREBP-1 or "E box"	6245-6251	ACACATG	7	Complement
41	SREBP-1 or "E box"	6288-6294	ACAACTG	7	Complement
42	SREBP-1 or "E box"	6623-6629	TCATTTG	7	Lead
43	SREBP-1 or "E box"	6836-6842	TCACCTG	7	Lead
44	SREBP-1 or "E box"	6837-6843	ACAGGTG	7	Complement
45	SREBP-1 or "E box"	7032-7038	ACAGGTG	7	Complement
46	SREBP-1 or "E box"	7069-7075	TCAGGTG	7	Lead
47	SREBP-1 or "E box"	7101-7107	ACATATG	7	Complement
48	SREBP-1 or "E box"	7138-7144	ACAGTTG	7	Lead
49	SREBP-1 or "E box"	7139-7145	TCAACTG	7	Complement
50	SREBP-1 or "E box"	7240-7246	ACACCTG	7	Complement
51	SREBP-1 or "E box"	7467-7473	ACAGGTG	7	Lead
52	SREBP-1 or "E box"	7640-7646	TCATTTG	7	Lead
53	SREBP-1 or "E box"	7641-7647	TCAAATG	7	Complement
54	SREBP-1 or "E box"	7653-7659	TCAGTTG	7	Lead
55	SREBP-1 or "E box"	7654-7660	ACAACTG	7	Complement
56	SREBP-1 or "E box"	7735-7741	ACAAATG	7	Lead
57	SREBP-1 or "E box"	7838-7844	TCAGGTG	7	Complement
58	SREBP-1 or "E box"	7880-7886	TCATCTG	7	Complement
59	SREBP-1 or "E box"	8051-8057	TCAGCTG	7	Lead
60	SREBP-1 or "E box"	8052-8058	TCAGCTG	7	Complement

Figure 16B